



Virginia Association of
**GENETIC
COUNSELORS**

15th Annual Virginia State Genetics Education Conference

**Thursday, Apr 29, 2021 – Friday April 30, 2021
Virtual Conference**

**We would like to thank our Platinum sponsors,
GeneDx and Invitae,
for their generous support of this year's meeting.**

This event has been submitted to the National Society of Genetic Counselors (NSGC) for approval of up to 1.3 Category 1 CEUs. (13 contact hours).

ATTENDEE INFORMATION

Registration:

<https://vaagc.vfairs.com/>

Thursday Apr 29, 2021 9:00 am – 4:30 pm

Friday April 30, 2021 9:00 am – 5:00 pm

Have Questions?

Click on the Information Desk in the Lobby

Conference Co-Chairs

Joyti Khokhar, MS, CGC, University of Virginia
VaAGC Western Region Representative

Brianna Murray, MS, CGC, Children's Hospital of the King's Daughters
VaAGC Eastern Region Representative

Conference Planning Committee

Dina Alaeddin, MGC, CGC, Inova Schar Cancer Institute

Wendi Betting, MS, CGC, University of Virginia

Allison Gossen, MS, CGC, University of Virginia

Melanie Hardy, MS, MS, CGC, JScreen at Emory University

Berkley Schmidt, MS, CGC, Virginia Commonwealth University

Adam Stucke, MS, CGC, Henrico Doctors Hospital Forest

Matthew Thomas, ScM, CGC, University of Virginia

Edward Williams, MS, CGC, Centra Health

Please complete the conference evaluation!

Link will be shared at the end of the conference.

THURSDAY Apr 29th - SPEAKERS

Speaker

Affiliation

Brianne Kirkpatrick, MS, LCGC

Watershed DNA

Kari Ring, MD, MS, FACOG

University of Virginia

Kayla Hamilton, MS, CGC

Dana-Farber Cancer Institute

Chelsea Chambers, MS, CGC

Virginia Commonwealth University

Swaroop Aradhya, PhD, FACMG

Invitae

Christina Peroutka, MD, FAAP, FACMG

University of Virginia

Jehannine Austin, PhD, FCAHS, CGC

University of British Columbia

Gretchen MacCarrick, MS, CGC

Johns Hopkins University

THURSDAY Apr 29th - AGENDA

Time	Title	Speaker(s)	CEU-Eligible Contact Hours
9:00 – 9:15 am	Conference Opening and Presidential Address	Joyti Khokhar, MS, CGC Brianna Murray, MS, CGC Lydia Higgs, MS, CGC	-
9:15 – 10:00 am	The Value of Genetic Testing and Genetic Counseling for Adopted Individuals	Brianne Kirkpatrick, MS, LCGC	0.75
10:00 – 10:45 am	Li-Fraumeni Syndrome: All that and a bag of CHIPs	Kari Ring, MD, MS, FACOG	0.75
10:45 – 11:00 am	15 Minute Break		
11:00 – 11:45 am	An Overview of Predisposition to Hematologic Malignancies	Kayla Hamilton, MS, CGC	0.75
11:45 – 12:30 pm	What Every Genetic Counselor Needs to Know about Neuromuscular and Neurodegenerative Diseases	Chelsea Chambers, MS, CGC	0.75
12:30 – 1:00 pm	Lunch Break		
1:00 – 1:45 pm	Not all Next Generation Sequencing is Equal: Advances in Variant Classification with RNA Analysis and Functional Protein Modeling	Swaroop Aradhya, PhD, FACMG	0.75
1:45 – 2:30 pm	Metabolic Disorders are Also Genetic! Expanding the Role of Genetic Counselors in Newborn Screening for Inborn Errors of Metabolism	Christina Peroutka, MD, FAAP, FACMG	0.75
2:30-2:45 pm	15 Minute Break		
2:45-3:45 pm	Evidence Based Genetic Counseling for Psychiatric Disorders	Jehannine Austin, PhD, FCAHS, CGC	1.0
3:45-4:30 pm	Of Mice and Men: What's New in Aortic Disease	Gretchen MacCarrick, MS, CGC	0.75
4:30 pm	Virtual Networking Social (optional)		

FRIDAY April 30th - SPEAKERS

Speaker	Affiliation
Erynn Gordon, MS, CGC	Genome Medical
Jane Juusola, PhD, FACMG	GeneDx
Ann Jewell, MS, LCGC	Virginia Commonwealth University
Tahnee Causey, MS, CGC	Virginia Commonwealth University MS in Genetic Counseling Program
Austin Bland, MS, CGC	Hawaii Department of Health, Minority Genetics Professional Network
Sarah Huguenard, MS, CGC	Baylor College of Medicine, Texas Society of Genetic Counselors
Smriti Singh, MSGC Candidate	Emory University, Genetic Counseling Trainee Platform for Racial Justice
Melanie Hardy, MS, MS, CGC (moderator)	JScreen at Emory University
Lauren Boucher, MSGC Candidate	Virginia Commonwealth University
Mariah Clark, MSGC Candidate	Virginia Commonwealth University
Aimee Macagney, MSGC Candidate	Virginia Commonwealth University
Jenna Miller, MS, CGC	Cooper Genomics
Marie Discenza, MS, CGC	Cooper Genomics
Emily Brown, MS, CGC	Johns Hopkins University
Aishwarya Arjunan, MS, MPH, CGC, CPH	Myriad Genetics

FRIDAY April 30th - AGENDA

Time	Title	Speaker(s)	CEU-Eligible Contact Hours
9:00 – 10:00 am	The Role of a GC in DTC Genetic Testing	Erynn Gordon, MS, CGC	1.0
10:00 – 10:45 am	Uncovering Answers with Exome/Genome Testing	Jane Juusola, PhD, FACMG	0.75
10:45 – 11:00 am	15 Minute Break		
11:00 – 11:30 am	Genetic Ophthalmology: The Eyes Have It!	Ann Jewell, MS, LCGC	0.50
11:30 am – 12:30 pm	DEIJ Efforts in the Genetic Counseling Profession – A Panel Discussion	Tahnee Causey, MS, CGC Austin Bland, MS, CGC Sarah Huguenard, MS, CGC Smriti Singh, MSGC Candidate Moderated by: Melanie Hardy, MS, MS, CGC	1.0
12:30 – 1:00 pm	Lunch Break		
1:00 – 1:20 pm	An Evaluation of Practices and Policies Utilized in Genetics Clinics Across the U.S. to Manage Referrals for Ehlers-Danlos Syndrome	Lauren Boucher, BS, MSGC Candidate	0.33
1:20 – 1:40 pm	Assessing Prenatal Genetic Counselors' Attitudes and Perceptions on Offering Abortion Options to Their Patients Amid Restrictive Legislation	Mariah Clark, BS, MSGC Candidate	0.33
1:40 – 2:00 pm	Assessing Perceived Productivity of Specialized and Multidisciplinary Genetic Counselors	Aimee Macagny, BS, MSGC Candidate	0.33
2:00 – 2:30 pm	Preimplantation Genetic Testing for Monogenic Disorders	Jenna Miller, MS, CGC Marie Discenza, MS, CGC	0.50
2:30 – 2:45 pm	15 Minute Break		
2:45 – 3:45 pm	Evaluation of Patients with Personal and Family Histories of Coronary Artery Disease	Emily Brown, CGC, MGC	1.0
3:45 – 4:45 pm	Carrier Screening & Systemic Racism in Genetic Testing	Aishwarya Arjunan, MS, MPH, CGC, CPH	1.0
4:45 – 5:00 pm	Conference Closing Remarks	Joyti Khokhar, MS, CGC Brianna Murray, MS, CGC	-

MEETING GOALS

Purpose:

To provide a venue for Virginia genetic counselors and other associated professionals to discuss current issues and topics in genetics, to learn about the variety of state programs and available resources, and to form a better network of communication.

Meeting Goals:

1. Attendees will be able to describe recent developments in the field of genetic counseling including diagnosis and management of genetic conditions.
2. Attendees will be able to identify genetics colleagues and representatives from companies and non-profit organizations in Virginia to aid in the provision of genetic counseling services.
3. Attendees will be able to apply new technologies, genetic testing and research results into the practice of genetic counseling.

Registration:

A registration fee of \$65 for members, \$90 for non-members, and \$45 for student members is due at the time of registration. For individuals requesting CEUs, an additional fee of \$25 will be required to be paid to the National Society of Genetic Counselors for the filing and awarding of CEU certificates. Space is limited.

WANT TO GET INVOLVED?

The VaAGC is always looking for **volunteers**.

If you would like to become more involved with the VaAGC, please sign up for one of the committees below at the registration desk or email the VaAGC at info@vaagc.com

- ◆ Bylaws and Public Policy Committee
- ◆ Website Design and Maintenance Committee
- ◆ State Meeting and Education Committee
 - ◆ Membership Committee
- ◆ Communication and Outreach Committee

VaAGC OFFICERS

THANK YOU to the 2020 – 2021 Officers:

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** : these designated 2020 – 2021 officers will be completing their terms effective at the close of this meeting

Meet your 2021 – 2022 Officers:

TBD	President
Lydia Higgs, MS, CGC, Carilion Clinic	Past-President
Jessica Watts, MS, CGC, GeneMatters	Secretary
TBD	Treasurer
TBD	Eastern Representative
Adam Stucke, Henrico Doctors Hospital Forest	Central Representative
TBD	Western Representative
Dina Alaeddin, MGC, CGC, Inova Schar Cancer Institute	Northern Representative
Kimberly Matthijssen, MS, CGC, Inova Schar Cancer Institute	Board of Directors
Hailey, Feldman, MS, CGC , Carilion Clinic	Board of Directors
TBD	Board of Directors

LEARNING OBJECTIVES

The participants will be able to:

- List the common reasons adoptees seek out genetic testing.
- Discuss the pros and cons of healthcare provider-mediated and directly-accessed genetic testing for adoptees.
- Identify external resources genetic counselors can use when supporting adopted clients and their families.
- Describe interpretation of germline testing for TP53 variants.
- Discuss clinical management of possible results.
- Identify indications for genetic counseling/testing for individuals with a personal or family history of a hematology malignancy.
- Explain risk assessment, differential diagnoses, and testing options for hematology malignancy indications.
- Discuss updates on new data regarding leukemia predisposition syndromes.
- Discuss the common neuromuscular and neurodegenerative disorders that could be seen in any Genetics clinic.
- Explain the current therapies and research available in neuromuscular diseases.
- Identify the genetic testing strategy and common concerns when counseling patients with neurodegenerative diseases.
- Explain the critical components of the NGS-based clinical genetic testing, including variant detection and variant interpretation.
- Summarize the concepts behind a typical variant interpretation framework and the categorization of applicable evidence.
- Identify the context for and benefits of RNA sequencing analysis in germline genetic testing as well as its limitations.
- Define the categories of variants whose clinical interpretation can be informed by evaluating effects on protein sequence.
- Discuss the history of the VA State Newborn Screen and future directions.
- Explain the role of genetic counseling in cystic fibrosis and hemoglobinopathy screens.
- Identify the critical role genetic counselors could fill in newborn screening for inborn errors of metabolism.
- Explain what is currently known about the genetics of psychiatric disorders.
- Discuss the importance of understanding causes of psychiatric illness for patients and their families.
- Discuss the outcomes of genetic counseling for psychiatric disorders with consideration of the implications for psychiatric genetic testing.
- Summarize advances in mouse model research applied to treatment of Vascular Ehlers Danlos syndrome.
- Discuss medical therapy trials in Marfan syndrome.
- Evaluate the rationale behind Familial thoracic aneurysm genes on clinically available gene panels.
- Describe differences between common forms of DTC testing.
- Compare the benefits and limitations of DTC genetic testing.
- Explain how to use DTC results that patients may present with in clinical practice.
- Describe the approach taken to analyze and interpret clinical genomics data, especially in the NICU/PICU setting.
- Discuss the reporting of clinically relevant variants that may be challenging to interrogate, including copy number variants (CNVs), mosaicism, mobile element insertions (MEIs), and variants in candidate genes.
- Explain why reanalyzing exome and genome data is important.
- Describe the most common indications for genetic ophthalmology referrals.
- Explain the team approach for genetic eye patients.
- Discuss genetic testing panels and results for inherited ocular disorders.
- Summarize the frontier of new gene therapies for the inherited retinopathies.
- Describe DELJ efforts in genetic counseling education and professional practice within Virginia and around the US.
- Discuss challenges, insights and successes in community-centered grassroots organizations and educational efforts in DELJ.
- Identify ways that genetic counselors can incorporate DELJ into their work, teaching, and supervision.
- Explain practices that are utilized in institutions across the U.S. to manage referrals for EDS.
- Discuss the motivations behind and impacts of the implementation of policies to manage EDS referrals.
- Explain how restrictive legislation surrounding abortion options has impacted prenatal genetic counselors' professional practice and their patients.
- Compare perceived productivity between specialized and multidisciplinary genetic counselors.
- Discuss the impact of various factors of on genetic counselors' perceived productivity.
- Define Preimplantation genetic testing (PGT) and its three subsets: PGT for Aneuploidy (PGT-A), PGT for Monogenic Disorders (PGT-M), and PGT for Structural Rearrangements (PGT-SR).
- Explain the utility, general process, and common challenges of PGT-M.
- Discuss an ethical issue common in preimplantation genetic testing for monogenic disorders.
- Recognize "red flags" in a personal and family history suggestive for a hereditary dyslipidemia.
- Describe utility of genetic testing for individuals within this patient population.
- Define the history of expanded carrier screening.
- Summarize how policies, guidelines, practice contribute to systemic racism and health disparities.

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