ISSUE
12
April 2023

Official Newsletter of the Pennsylvania Association of Genetic Counselors

PAGC News

The PAGC Newsletter is brought to you by members of the PAGC Membership Committee

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If you have anything that you would like to share including upcoming events, seminars, a recent publication, an interesting book, movie, or podcast, etc., please contact us at : PAGCmembership@gmail.com

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www.pennsylvaniagc.org

PAGC held the 7th Annual Conference March 3-4, 2023 at Geisinger Medical Center

What a great experience it was to be able to connect with colleagues in person this year for the annual conference! The PAGC Education Committee met the challenge of providing quality presentations for in-person and virtual attendees. There was a total of 141 attendees this year, including 27 GC students, plus 27 speakers; 109 genetic counselors obtained CEUs.

All around – a successful, engaging event. We look forward to next year!

Please visit the PAGC website for post-conference materials: Information on the Warren Alpert Foundation grant to increase diversity in genetic counseling student enrollment and updated management guidelines for 22q11.2 Deletion syndrome.

Continue reading for **topic highlights** from the conference.

Run to the Light
By Laura King Edwards

Book Review by Emily Lancaster, MS, CGC

When Laura's sister Taylor was diagnosed with Batten disease*, Laura and their mother created the charity Taylor's Tale to fund research towards a cure for this fatal genetic condition. Laura and her mother started attending conferences about Batten disease where they would forgo the family support sessions, something many genetic counselors might cringe at, and instead dove in on the hard science talks and arranged meetings directly with the researchers who might potentially find a treatment. Laura and her mother were driven to do everything they could to understand how Batten disease works and ways to fight back against the disease that was starting to affect Taylor more and more. Meanwhile, when Taylor's vision progressed to the point of blindness, she decided to run a 5k while using a sighted guide, which in turn inspired Laura to start running marathons to help raise awareness of her sister's story. This personal challenge ultimately culminated in Laura running a race while blindfolded with a guide in a mirror of Taylor's run, allowing her to run miles in her sister's shoes.

*Also known as neuronal ceroid lipofuscinosis

Conference Topic Highlight:

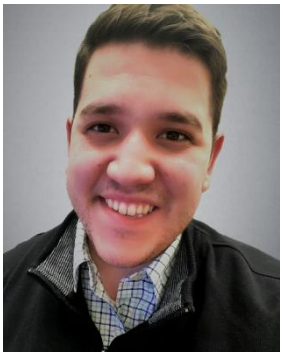
Genetic Counselors in Academic Research Roles

Summarized by Emily Lancaster, MS, CGC

Panel Participants:
Zachary Salvati, MS, CGC (Geisinger)
Nadene Henderson MS, CGC (Pitt)
Elizabeth McCormick MS, CGC (CHOP)
Moderator: Margaret Harr MS, CGC (CHOP)

The 2022 NSGC PSS showed that 50% of genetic counselors are involved in research activities and 61% have published in the last 2 years, so it's timely that this talk highlighted what a role outside of the clinic can look like and how clinical skills can be utilized in academic research. Both roles can involve discussing results with patients, reviewing potential treatment options, consenting for research enrollment, reviewing patient charts for clinical history, assessing potential inheritance patterns, and touching base with patients and families on how they are coping. An especially important skill for research studies that was highlighted is a genetic counselor's ability to ask semi-structured questions during interviews, something not all medical professionals and PhDs are taught but is valuable for information gathering. Some recommended resources for individuals who want to learn more about involvement in research include the NSGC Research SIG and NSGC RQO Committee; certification as a research coordinator can be pursued for those looking for a more active role in research involvement. This panel talk also provided encouragement to present posters and case reports and share important information about cases, which can be done at national conferences such as NSGC, ACMG, or ASHG, or by publishing in a journal. While the Journal of Genetic Counseling is the first thought for many on where to publish, it was also discussed to branch out and submit to other journals as well, which will help get our names and profession into the awareness of the scientific community.

Featured Genetic Counselor: **Zachary Salvati**



Genetic Counseling Program: University of Oklahoma
Year of graduation: 2018

Current Employer: Geisinger Medical Center

What attracted you to pursue a career in genetic counseling?
I've always had a strong desire to help others. Growing up outside Chicago in the '90s, I told people I would be a "doctor in the NBA" because everything revolved around MJ and the Bulls. Even from a young age I knew my place was somewhere in the medical field!

Fast forward to the genetics unit in my first high school biology course – This was one of the first times I felt fully engaged in the sciences. I researched possible careers that incorporated both genetics and seeing patients, and that's when I found out about the genetic counseling profession. Shadowing opportunities and a summer internship in undergrad gave me multiple chances to test the waters before applying to grad schools and diving headfirst into this career.

What are your responsibilities in your current position?
My responsibilities are primarily related to research coordination since I am a genetic counselor with 1.0 FTE across different research studies. My day-to-day varies depending on the project, but it can include provider and/or patient interviews, chart reviews, collecting and analyzing data sets, and helping disseminate information via local organizational networks and/or through conference abstracts and manuscript writing. I enjoy qualitative research methods, and I help lead Geisinger's Qualitative Research Multidisciplinary Meeting, providing a peer-to-peer platform to share resources and research goals.

Any patient contact I have is currently done through the lens of research, like returning results from Multi-Cancer Early Detection screening that looks for evidence of early-stage cancer in any otherwise-healthy cohort of patient-participants. I advocate that genetic counselors are well-equipped to lead projects with sensitive subject matter due to our training and ability to maintain contact/coordinate care.

How have you seen opportunities for genetic counselors evolve during the course of your career? Is there a pivotal occurrence that opened a career opportunity for you?
This is a great question. Geisinger has had a larger team of genetic counselors for many years. When I started ~5 years ago, there weren't many GCs fully dedicated to research but almost everyone had split clinic/research time. The latter is still true, but there's a growing need for GCs to not only be a part of research but to lead these projects. I transitioned from split clinic/research time to fully research back in 2019, receiving both on-the-job and formal training to foster growth in my own career path and bolster support for other GCs looking to get involved in research. This career path has allowed me to continue my education through implementation of science coursework and actively participating in the GC research community (e.g., I chair the NSGC Research SIG, and I am a member of the NSGC Research, Quality, and Outcomes committee). Switching to 1.0 FTE in research and having the ability to focus on the research subspecialty here at Geisinger opened many doors for me. I'm grateful to have those opportunities!

What "I wish I knew then what I know now" advice would you give to recent GC grads?
Take things one step at a time and (try to) appreciate growth. You're not going to be the best version of your GC-self right out of grad school, but you know more than you think you do! Imposter syndrome is rampant across most professions and genetic counseling isn't unique in that respect. Humility is a lauded quality, understandably, but for me it can be easier to put more weight on my failures than my accomplishments. You will make mistakes, but you will also succeed! Find a mentor and a community that will keep you accountable and celebrate with you, and don't be so hard on yourself. The best is yet to come!

Conference Topic Highlight:

Genetic Testing of Circulating Tumor DNA

Presenter:
Natalie Carter MS, CGC (Guardant Health)

Summarized by Amy Kunz, MS, CGC

At its core, all cancer is a genetic disease – caused by a series of mutations that occur within a cell's DNA. The emerging technology discussed in this talk takes advantage of the fact that tumors shed DNA into the bloodstream via apoptosis and necrosis, collectively known as circulating tumor DNA (ctDNA). Amazingly, often ctDNA can even detect cancer prior to our ability to detect cancer via imaging. Currently, in oncology, ctDNA is mostly used in the advanced disease setting to select patient candidates for targeted therapies, of which there are many. Targeted therapy is in contrast to standard chemotherapy, with the goal of prolonging disease remission in patients. The most exciting aspects of ctDNA are in its future applications. These include monitoring the tumor's response to therapy and better determining who out of those with early-stage disease is more likely to recur (i.e., the ability to detect microscopic residual disease, or MRD). Much attention has also been applied to the ability to screen for early-stage cancers using ctDNA technology. Given the rapid advancements in this area of cancer treatment, genetic counselors are well poised to be part of the integration of these new tests into oncology practices.

Conference Topic Highlight:

Little a Advocacy

Presenter:
Rebecca Mueller PhD, CGC
University of Pennsylvania
Department of Medical Ethics & Health Policy

Summarized by Susan Walther, MS, CGC

Advocacy involves promoting the interests or cause of a group of people and helping them find their voice while effecting positive change. 'Little a' represents advocacy on the micro-level. Each GC holds responsibility for improving patient access to healthcare, and, specifically, access to appropriate genetic services. This responsibility is especially powerful for patient populations who routinely feel marginalized by our healthcare system and insurance payers.

Areas that genetic counselors can consider when addressing JEDI issues include:

- (CLINIC): You can't manage what you can't measure – use EMR to get the numbers for types of referrals, frequency of referrals based on groups of patient types, and equity of access to genetic services (consider follow up rate, insurance type, healthy literacy). Address with patients why information is requested in a genetic counseling session, and how information provided by the patient is used in health management.

- (RESEARCH & TEACHING): Question the expertise needed to educate on race/ancestral/ethnic background and cultural differences. Over-emphasizing past history can miss current issues, and making broad assumptions can result in failing to identify an issue of importance to a specific patient (e.g., assuming that Tuskegee experiments still limit research participation for people of color). We classify people all the time, but think about the reasons and consequences. To find solutions for diverse stakeholders in healthcare, the needs at different levels must be recognized and acknowledged – patients, clinicians, and administration of the hospital/institution.

Conference Topic Highlight:

Prenatal Genetic Counseling in Pennsylvania after *Dobbs v. Jackson Women's Health Organization*

Presenter:
Rebecca Belles MS, CGC
Geisinger Health System

Summarized by Susan Walther, MS, CGC

In *Roe* v. *Wade*, the case decided by the U.S. Supreme Court in January 1973, seven of the nine justices agreed that the due process clause of the 14th Amendment — which says that no state shall "deprive any person of life, liberty, or property, without due process of law" — implies a right to privacy when choosing to end a pregnancy through abortion. In June 2022, *Roe* v. *Wade* was overturned with the court majority deciding that the U.S. Constitution does not confer a right to abortion, and, therefore, abortion is a matter to be decided by states and the voters in the states. *Dobbs* v. *Jackson Women's Health Organization*, the Mississippi case that was the basis for the court's overturn decision, banned abortion after 15 weeks and applied penalties to abortion providers. Within weeks following the Supreme Court's ruling, several states reverted to previous abortion ban laws that had been on their books before *Roe* v. *Wade* was decided. By June 2022, 13 states had passed trigger laws that were designed to outlaw most abortions if the Supreme Court threw out the constitutional right to end a pregnancy. Currently, Pennsylvania does not have a trigger law, but Pennsylvania is considered a hostile state (Guttmacher classification) because there is no state constitutional law(s) to protect access to abortion. In Pennsylvania, bills can be introduced in the General Assembly, and if approved in the house and senate, and approved in two back-to-back assemblies, then a vote can be taken to voters, bypassing the governor. Currently, in Pennsylvania, abortion is legal up to 24 weeks after a person's last menstrual period. A patient seeking an abortion must receive state-mandated counseling that includes information designed to discourage them from having an abortion, and then wait 24 hours before the procedure is provided. Health plans offered in the state's health exchange under the Affordable Care Act can only cover abortion if the person's life is endangered, or in cases of rape or incest. If Pennsylvania's current abortion laws remain intact, experts say there may be another outcome in a post-*Roe* world: Abortion providers in the state will likely see a surge in demand from out-of-state patients seeking the procedure.

Online resources:

- Guttmacher.org – tracking state bills
- reproductiverights.org – what is currently happening, maps available
- abortionfunds.org/state-legislative-updates
- Medical students for choice (msfc.org)
- GenuineCollective.org – genetics providers united in efforts for reproductive justice

GC STUDENTS: PAGC features student profiles on the website. Please contact pagcmembership@gmail.com for the list of questions. The profile questions are best-suited for second year students. A headshot is also needed for the profile. You can view student profiles at the bottom of the homepage of the website: https://www.pennsylvaniagc.org/

Conference Topic Highlight:

Nephrology Genetics

Presenters:
Alex Change, MD
Gretchen Urban MS, CGC
Geisinger Health System

Summarized by Kelsey Bohnert, MS, CGC

Nearly 37 million Americans have kidney disease, where 10-15% of diseases are attributed to genetics, and of those, 30% report a family history of disease. While most diseases are adult-onset, a significant number of children have chronic kidney disease. A genetics referral is often made based on family history, current clinical picture, including biopsy, transplant evaluation, or clinical suspicion. Genetic testing can clarify the clinical picture, such as reclassify a diagnosis or inform prognosis; target management, such as extra-renal intervention, possible therapies, eligibility for clinical trials, and consideration for transplant; and for at-risk family member considerations. The approach to genetic testing for kidney disease is varied and is largely based on clinical presentation and family history. In some cases, a targeted approach using single gene testing or a panel may be appropriate, whereas in other cases, whole exome or genome sequencing should be considered. There are some disorders that may require molecular diagnosis using non-specific testing such chromosome analysis, MLPA, PCR, or tandem repeat. In all cases, it is important to consider the limitations of the technology being used—for example PKD1, the gene most linked with autosomal dominant polycystic kidney disease (ADPKD), is difficult to sequence and, thus, not included on all panels, including cystic kidney panels.

The NSGC Renal SIG has put together a helpful tool to identify patients that might benefit from a renal genetic evaluation. It can be accessed here.

Additionally, the following articles offer helpful insights into the care of patients with renal disease:

- Clinical impact of genomic testing in patients with suspected monogenic kidney disease by Jayasinghe et al: Discusses a diagnostic yield of exome (39%) in a cohort of 204 patients with renal disease, along with the factors contributing to a higher diagnostic yield.
- Vignette-Based Reflections to Inform Genetic Testing Policies in Living Kidney Donors by Gurmukteshwar et al.: Highlights several case studies of how genetic testing should be integrated into transplant testing donor evaluation.
- The Clinical Impact of Genetic Testing in Outpatient General and Transplant Nephrology: A Tertiary Centre Experience by Sankar et al: Abstract submitted to the American Society of Nephrology for Kidney week 2022 highlighting the diagnostic yield of genetic testing within the Geisinger Healthcare System.
- Novel genetic testing model: A collaboration between genetic counselors and nephrology by Amilie-Wolf et al.: Introduces a consult-based model where genetic counselors work with nephrologists as part of a utilization management effort to increase access to genetic testing.

PAGC Committees

Volunteers are always welcome!

Contact committee chair if you are interested in being involved

Education

Chairs:
Amanda Back (backa@chop.edu))
Shannon Terek (terks1@chop.edu)

- Plan and implement the annual conference
- Manage CEU submission to NSGC

Professional Issues

Chair: Becky Sullenberger (becpitt30@gmail.com)

- Work to update GC licensure in PA
- Examine barriers to credentialing of GCs in PA
- Develop education on process & benefits of credentialing

Genetic Services

Chair: Gabby Shermanski (gtshermanski@geisinger.edu)

- Working on Pennsylvania Professional Status Survey
- Evaluate GC services in PA

Membership

Chair: Susan Walther (susanwalther1203@gmail.com)

- Implement website design and maintain content
- Manage e-blast communications/facilitate conference registration
- Develop content for PAGC newsletter

Justice, Equity, Diversity and Inclusion

Co-Chairs: Kelsey Bohnert (kelsey.bohnert@chp.edu) and
Aaron Baldwin (aaron.baldwin@pennmedicine.upenn.edu)

Call for JEDI speakers! The PAGC JEDI committee is currently working on a CEU series addressing different aspects of JEDI in the genetic counseling profession including clinical supervision, underrepresented groups, patient experience, research, pathway to the profession and mentorship. If you're interested in being a speaker during this series, please reach out to the JEDI co-chairs.