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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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Emily Lancaster, MS, CGC

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If you have anything that you would like to share, including upcoming events, seminars, an exciting new career role, etc., please contact us at : PAGCmembership@gmail.com

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PAGC
Pennsylvania Association
of Genetic Counselors

PAGC ANNUAL CONFERENCE
SAVE THE DATE
SEE PAGE 7

PENNSYLVANIA PROFESSIONAL STATUS SURNEY

Pennsylvania Association of Genetic Counselors (PAGC)

2023 Professional Status Survey (PSS) Summary

55% Response Rate from PAGC Members
+ 25 non-member responses

Most GC respondents are employed full time in a salaried position.
Median Salary Range: $90,000 - $109,999
Majority Range: $70,000 - $109,999

Additional **professional activities** include:

- Student supervision
- Lecturing
- Involvement with GC graduate programs
- Research
- Volunteer in professional organization(s), patient advocacy organization or community
- Faculty position

Of respondents who reported providing direct patient care, **97%** see at least some patients in a telegenetics setting.

The full PAGC 2023 PSS Report will be available to members in mid-October 2024.

PAGC
Pennsylvania Association of Genetic Counselors
https://www.pennsylvaniagc.org

Summaries of Presentations from the 2024 PAGC Annual Conference

Pages 2,4,5,6

Genetic Counseling Clinical Updates: Cardio Genetics

Summarized by Amy Kunz

Kristy DiLoreto kicked off the first session of the 2024 spring conference with sharing clinical updates in the area of cardiology. When surveyed, more than half of genetic counselors practicing in the cardio genetics space feel that the current traditional service delivery model is insufficient in caring for patients. As with nearly all subspecialties, there is a high demand for testing; however, per a 2024 study reviewing insurance claims data, only 1.1% of individuals with cardiomyopathy underwent genetic testing. A number of models have been successfully implemented to help close this gap in access including non-genetics provider ordering tests, genetic counselors embedded in non-genetics cardiology clinics, and synchronous encounters with cardiologists. A number of emerging treatments for hereditary cardiovascular syndromes has further highlighted the importance of genetic testing, including therapies for hereditary transthyretin amyloidosis (hATTR) and symptomatic obstructive hypertrophic cardiomyopathy. This talk highlighted resources for genetic counselors who see patients with heritable cardiac phenotypes - the Cardiogenomic Guidelines Database, the Cardiogenomic Testing Alliance, and Genetic Cardiomyopathy Awareness Consortium.

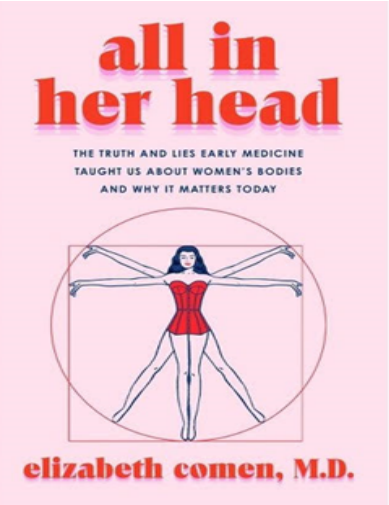
Technology AI Use in Genetic Counseling: Perspectives from the Genetic Counselor and the Lab

Speakers: Becky Milewski, MS, CGC and Jeffery Bissonnette, MS, CGC

Summarized by Emily Lancaster

Artificial Intelligence (AI) refers to a simulation/approximation of human intelligence in machines with goals of computer-enhanced learning, reasoning, and perception. Currently some labs are using AI methods to assist in variant interpretation and triaging to determine if genes/variants of interest meet reporting requirements. **Emedgene** is one such system to help determine variants that are “candidates” for a proband’s phenotype. In Meng et al. 2023 (PMID 36939041) they reported in 180 retrospective solved cases, 96.7% had the reportable finding in the top 10 variants called by Emedgene. For 334 prospective cases, 28.7% were considered solved, with an additional 12.6% having a possible diagnosis – reported lab solve rates can be in the 26-30% range. One advantage was that Emedgene has a faster turnaround time (TAT) and as exome and genome sequencing are being used more for patient cases, and TAT is a crucial hold up, use of AI can lead to increase in lab volume and more access for care. However, there are limitations, including inability to assess certain variant types, decreased utility for patients with multiple diagnoses, hypomorphic alleles, and the ever-changing nature of genetics with new gene/disease relationships, new clinical features, and population databases being updated. For literature in variant interpretation, AI can help reduce the manual workflow, although it is not a replacement for expert review, as publications may use different nomenclature or disease terms that can cause problems for AI, along with formatting issues, such as tables and supplemental materials being more difficult for AI to identify listed variants. When used effectively, AI can remove/deprioritize likely false positives and increase specificity, prioritize publications for review based on potential evidence and section of the publication for a manual review, and help resolve nomenclature issues/discrepancies.

All in Her Head: The Truth and Lies Early Medicine Taught Us About Women's Bodies and Why It Matters Today



By Elizabeth Comen, M.D.

Book Review by Amy Kunz, MS, CGC

The book delves into the biases and misconceptions that have plagued women's healthcare for centuries, revealing how these issues continue to affect women today. As an oncologist and medical historian, Comen sheds light on the systemic problems that have led to the marginalization of women's health concerns and draws parallels to present day anecdotes with her own female patients.

Systematized by organ system, Comen traces the roots of medical misogyny back to early medical practices, highlighting how women's bodies were often misunderstood and mistreated. One theme is that male-dominated medical institutions have historically viewed women's bodies as flawed versions of the male ideal. I learned for example that women's colons are longer than men's, making colonoscopies more time consuming and difficult.

Another unsurprising theme is how a woman's symptoms are often thought to be of psychological origin rather than of true pathology. "Conditions ranging from heart attack to rhythm abnormalities were broadly dismissed in female patients as a symptom of emotional unbalance, rather than organic circulatory disease."

One of the more interesting connections that Comen describes is how certain illnesses were seen as ideal in the female form because they enhanced one's attractiveness or qualities as a wife, values of which were of most importance at their time. "The look of tuberculosis did tend to align with the beauty standards of the time: skin so pale it became transparent, emaciation brought on by lack of appetite, a natural blush in the cheeks and lips."

The misogyny extends to blaming women for the health scourges on society that victimize men. Beyond the more well-known Typhoid Mary, we learn of Dr. Thomas Parran (of Tuskegee Syphilis Study infamy) and the American Plan. In this episode, women were being treated as "scapegoats for contagion — and a system that perpetuates said scapegoating not only in medical settings, but in larger society, often with the authoritarian force of the government's public health apparatus behind it."

Comen's writing is both informative and engaging, dryly humorous and deeply empathetic. This approach not only educates but also empowers readers to advocate for better healthcare for women. The author discusses how modern medical practices still reflect outdated beliefs and biases, leading to inadequate treatment. Overall, All in Her Head is a must-read for anyone interested in women's health and medical history - "a cautionary tale about how easy it is for mistaken beliefs to become entrenched in medicine, and how difficult they are to correct."

Panel Discussion: Genetic Counseling in Advocacy

Panel Participants:
Andrea Durst DrPH, MS, CGC (University of Pittsburgh)
Samantha Sandy MS, CGC
Kellyn Madden MS, CGC (Friedreich's Ataxia Research Alliance)

Moderator: Dana Farengo Clark MS, CGC (University of Pennsylvania)

Summarized by Emily Lancaster, MS, CGC

Advocacy is "an activity by an individual or group that aims to influence decisions within political, economic, and social institutions", and from ASHG, "Scientists as advocates bring expertise in evidence-based decision making and, by reaching out to policymakers, we can help influence them to pursue policies that advance biomedical science and the appropriate application of genetic in health and society." Genetic counselors, specifically, can advocate for patients, the field of genetic counseling, and ourselves in a myriad of ways.

For Public Health Advocacy, **Dr. Andrea Durst** talked about patient advocacy, provider education, systems advocacy, and self-advocacy under the umbrella of public health – focusing on people and communities that can extend beyond solely health to also include other aspects, including work and play. Public Health Genetics Week is May 20-24, and more information can be found at https://phgw.org. The NYMAC Regional Genetics Network offers support for regional infrastructure, provides education-related activities, and facilitates the use of telehealth/telemedicine and has multidisciplinary teams with reps across Pennsylvania – they developed "Why Genetics" infographics that are available in several languages.

Samantha Sandy discussed the Payer Space and advocacy and how genetic counselors are involved in utilization management, patient liaison, policy development, claims review, education, contracts/pricing, vendor management, and fraud/waste/abuse data analytics. Different payors can have incongruent policies, such as restrictions on gene panel composition, regional influences, and different state Medicaid coverage decisions which lead to decreased access to services. Genetic counselors' involvement helps ensure patients are getting the right high quality genetic tests along with development of policies leading to standardization across payers.

Kellyn Madden discussed Patient Advocacy within the Friedrich's Ataxia Research Alliance and outlined the importance of the patient voice during the process of obtaining FDA approval for Skyclarys (omaveloxelone). Within a patient advocacy organization, genetic counselors can center the patient experience, help patient advocates hone their voice, advocate on behalf of the patient community, support patients with their advocacy, help to build relationships with stakeholders, and tailor education for the patient community and biopharma/researchers/regulatory agencies.

Ultimately, there are many ways for a genetic counselor to be involved in advocacy depending on their interests and position.

Genetic Counseling Clinical Updates: Post Mortem Genetics

Summarized by Amy Kunz

There can be tremendously helpful information gained from genetic testing in those family members who have passed from a suspected cardiac genetic condition, however the coordination of such genetic testing in the post mortem setting has unique challenges, as described by **Victoria Bacon** and **Jessica Sebastian**. Cost and insurance coverage for such situations can be particularly burdensome as insurance coverage typically ends after death. Samples are not guaranteed to be available, and the coordination of testing also potentially requires many more cooperative parties and conversations around consent – all of which are complicated by the emotional complexities of loss of a family member, and even more so when the loss involves an infant. Case examples illustrated how when all the right pieces fall into place, these efforts can yield significant meaning to families seeking closure.

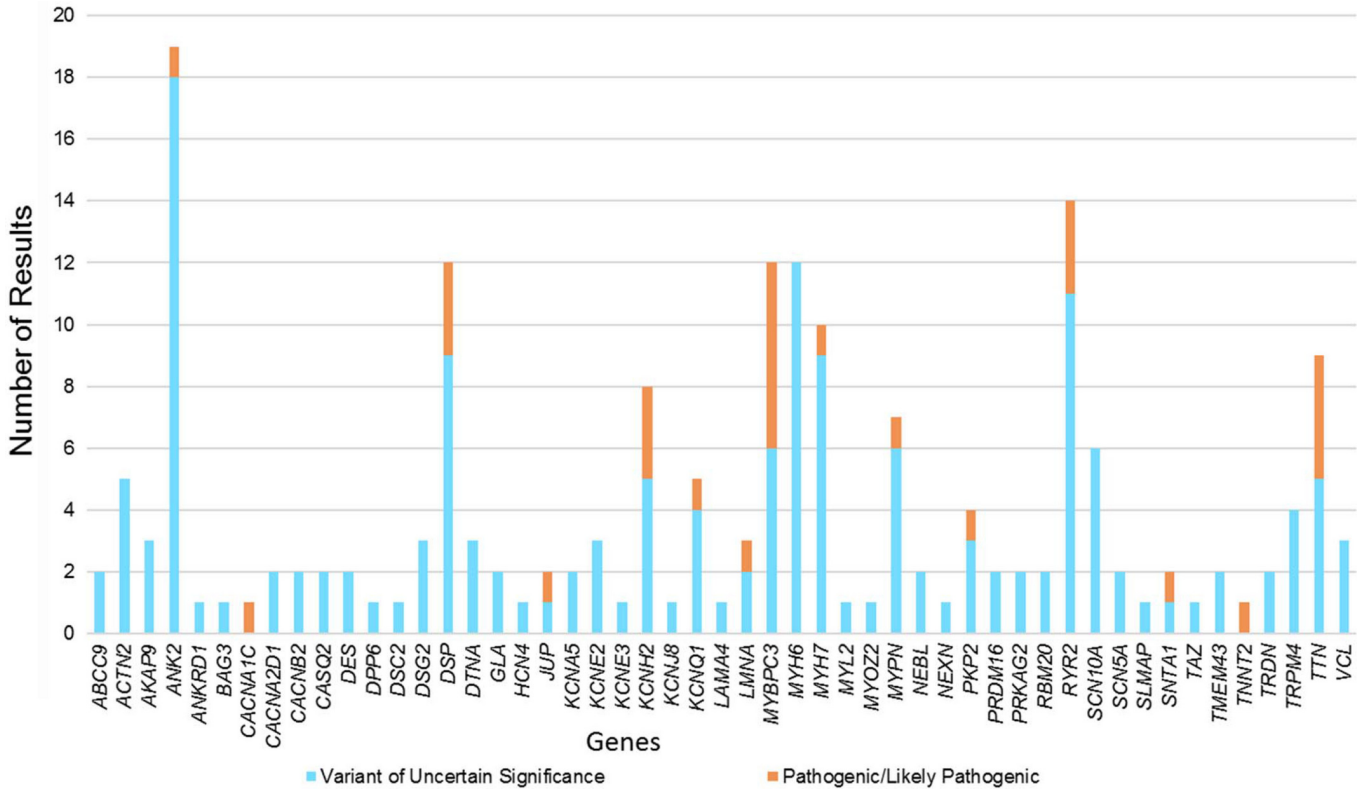
Additional Cardiac Post Mortem Info:

Lessons learned from testing cardiac channelopathy and cardiomyopathy genes in individuals who died suddenly: A two-year prospective study in a large medical examiner's office with an in-house molecular genetics laboratory and genetic counseling services

Nori Williams, Elizabeth Manderski, Sarah Stewart, Ruijun Bao, Yingying Tang

J Genet Couns 2020 Apr;29(2):293-302

https://doi.org/10.1002/jgc4.1157



Panel Discussion: Genetic Counseling in Population Screening

Summarized by Amy Kunz

Population screening remains one of the most discussed hot topics in genetic counseling. During this panel lessons were learned from the efforts of **Megan Betts** of Wellspan Health, a community hospital system that partnered with an industry leader, Helix, to integrate genomics into the healthcare organization.

Juliann Savatt took us through the history of MyCode, Geisinger Health System's biobank that has enrolled nearly a quarter of a million individuals and has delivered actionable genomic results to more than 5000 recipients – the majority of whom were unaware of their genomic risk.

Finally, we heard from **Dr. Mylynda Massart**, a primary care physician who provides care throughout a patient's lifetime and utilizes genomics in implementation of preventative care. A framework for how to make population screening a reality was at the forefront of discussion including social and ethical implications and the likely strain on health systems. This panel posed the challenge to its audience: Stop asking "Are we ready?" and ask "How do we get ready?"

Assessing Patient Perceptions and Understandings of Genetic Testing After Recurrent Pregnancy Loss (Poster)

Emily Hrach, BS; Svetlana A. Yatsenko, MD, Robin E. Grubs, MS, PhD, LCGC ; Elizabeth Sheehan, MS, CGC; Jenna Carlson, PhD

Genetic testing can help to provide an explanation for many couples experiencing recurrent pregnancy loss (RPL). However, there is limited data regarding the experiences of RPL patients undergoing genetic testing. This study assessed patient experiences, perceptions, and understandings of genetic testing for RPL. An online questionnaire was developed and distributed in relevant clinics as well as through social media and support groups. In total, 115 respondents met inclusion criteria, 105 completed the survey, and all responses were analyzed. Despite ACOG and ASRM practice recommendations, **only 63.7% of RPL patients were offered genetic testing on the products of conception (POC)**. Furthermore, only 67.9% and 54.5% were offered genetic testing for themselves and their partners, respectively. Respondents who reported that they were offered genetic testing but did not complete it cited cost, emotional stress, previous genetic screening, and logistical issues as reasons why they did not complete genetic testing on the POC or themselves/their partners. Overall, respondents recognized the potential of genetic testing to provide explanations for RPL and help plan for future pregnancies; **93.52% indicated that they would do/have done genetic testing to find an explanation for a miscarriage**. Respondents who found an explanation for RPL through genetic testing were more likely to recognize the utility and limitations of genetic testing than those who did not do genetic testing or who received uninformative/negative results. Outcomes of genetic testing did not significantly impact respondents' likelihood of utilizing pre-implantation genetic screening, additional screenings during pregnancy, egg/sperm donation, or adoption. Finally, 20.4% of respondents indicated that they would blame themselves if they were found to have a genetic change that explained their history of miscarriage, while only 8.3% indicated that they would blame their partner. The findings of this study indicate the need for additional patient education and resources regarding the risks, benefits, and limitations of genetic testing for RPL as well as the need for systematic implementation of current guidelines into clinical practice.

PAGC Committees

Volunteers are always welcome!

Contact committee chair if you are interested in being involved

Education

Chairs:
Lucy Galea (lucygalea@gmail.com)
Shannon Terek (tereks1@chop.edu)

Planning for the annual conference is underway for speakers, events, sponsors, and vendors. Please contact the committee chair to be involved in the planning, abstract review, or volunteering for on-site logistics.

SAVE THE DATES: Annual Conference
April 3-4, 2025 in Philadelphia (location TBD)

Professional Issues

Chair: Becky Belles (rsbelles@geisinger.edu)

- Work to update GC licensure in PA
- Examine barriers to credentialing of GCs in PA
- Create awareness of healthcare bills being considered in PA legislature

Genetic Services

Chair: Gabby Shermanski (gtshermanski@geisinger.edu)

- Design and implement Pennsylvania Professional Status Survey
- Evaluate and promote GC services in PA
- Create social media content

Membership

Chair: Susan Walther (susanwalther1203@gmail.com)

- Maintain PAGC website content
- Create e-blast communications
- Manage registration for annual conference
- Develop articles for PAGC newsletter

Justice, Equity, Diversity and Inclusion

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

This committee will be working on developing a recorded webinar series, as well as partnering with high schools and genetic counseling programs to increase high school students' exposure to the field of genetic counseling through the creation of a toolkit. Please contact the committee chairs for more information or to express your interest in joining the committee.