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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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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 Website:
www.pennsylvaniagc.org

ANNOUNCEMENT

The Executive Board is pleased to announce that the amendments to the PAGC bylaws, including the addition of the Education and JEDI committees, have been approved by the NSGC Board of Directors.

MEMBERSHIP

PAGC has grown our membership to 93 Full Members, 5 Associate Members, and 21 Student Members. And there were 119 people who registered for the 2022 annual conference. Thank you for your ongoing support in growing our organization!

Please note: PAGC operates under the umbrella of NSGC for our non-profit status. Due to NSGC bylaws, PAGC memberships must renew each calendar year and be in effect from January to December. When PAGC transitioned to our new website in 2021, memberships were set up to automatically renew based on a 12-month period from the date of initial purchase. **The automatic renewal was an error, and the Membership Committee is starting to cancel memberships that are under the automatic renewal process**. If you purchased a separate membership for 2022, it is still valid until December. You might receive a cancellation notification. Please feel free to email PAGCmembership @gmail.com with any questions or concerns.

Enjoy the benefits of being a member of PAGC, including discounted registration fees for the annual Spring conference, access to the newsletter, and committee opportunities for professional development (*see page 6*).

Check out the "Find A Genetic Counselor" Feature on the PennsylvaniaGC.org website! The feature now includes a Google map of genetic service locations. Let the membership Committee know if a clinical site needs to be added or information updated.

https://www.pennsylvaniagc.org/patients-and-providers

The Boy Who Loved Too Much: A True Story of Pathological Friendliness

By Jennifer Latson

Review by Emily Lancaster, MS, CGC

This book tells the true story of Eli, a 12-year-old boy with Williams syndrome, and his mother Gayle. Gayle has been a single mother and Eli's biggest advocate for years, keeping him as happy and safe as she can. Gayle shares with Jennifer, the author, what it was like to go through receiving Eli's diagnosis, advocating for Eli to get him the right services and therapies, and her fears as he enters adolescence with what might come next. At 12-years-old, Eli is entering the stage of his life where he wants more independence and tells his mom he wants a girlfriend; these are things that Gayle also wants for him while wanting to keep him from getting hurt, and that balance can be especially hard for parents of children with Williams syndrome. This book offers a truly personal look into their daily lives and does a wonderful job showing how hard it can be, especially for a single parent, to raise a child with a genetic condition while also showing how much joy and love Gayle and Eli have in their lives.

Featured Genetic Counselor: Jacqueline Hoover
Genetic Counseling Program: Sarah Lawrence College
Year of graduation: 1999
Current Employer: Allegheny Health Network

What attracted you to pursue a career in genetic counseling?
While doing an externship during my junior year of college with a gynecologist in a hospital setting, he introduced me to a genetic counselor who saw prenatal patients. I was astounded that there was a career that combined my love of the medical field, teaching, working with patients and my obsession with anything genetics! At the time, there was not much information about the career, and I did a lot of the career research on my own. I have also, through the years, loved the fact that I continue to learn in an ever changing and evolving field.

What are your responsibilities in your current position?
I am a full-time cancer genetic counselor with Allegheny Health Network, and I see patients both in person as well as remotely. In addition to risk assessment and coordinating hereditary germline testing, I serve on the molecular tumor board. I also work with students who rotate in our department to arrange their schedules, as well as provide feedback and grading. The greatest reward of having been a genetic counselor for so many years is being able to help train the genetic counselors of the future. In my spare time, I am a PIAA volleyball official and love listening to true crime podcasts.

How has COVID-19 affected your job responsibilities?
With the outbreak of COVID and moving to fully remote consultations, I feel that I needed to refine my abilities to assess patients' understanding in new ways. Without having the ability to judge non-verbal communication, I learned to rely on asking open-ended questions to generate more conversation on their end. This method not only allowed me to judge their understanding but also better determine underlying motivations for testing and further discussions on relevance specifically to them and their families.

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Featured Genetic Counselor: Jacqueline Hoover

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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?
The most striking change in genetic counseling over the course of my career has been the specialization into areas of practice. When I first started seeing patients in 1999, I saw prenatal, pediatric, and cancer patients for genetic counseling, depending on the day of the week. Now, it would be very difficult to stay up to date on new information without finding a specialty. When I first moved back to Pennsylvania after working in New York for 5 years, I was at Children's Hospital of Pittsburgh doing pediatrics for almost 10 years but knew I ultimately would love to get back into oncology genetic counseling. My opportunity came in 2013/2014 with the launch of hereditary cancer panels. I went to work for 1 year for a testing laboratory, which allowed me time to become up to date with the new hereditary cancer predisposition genes, prior to starting to see patients in my current position with Allegheny Health Network.

What "I wish I knew then what I know now" advice would you give to recent GC grads?
Most genetic counselors I know, including myself, are very detail-oriented. We also want to do our jobs perfectly and not make mistakes or to not know the answers. But I would argue that sometimes what feels like our biggest challenges, or even defeats, have the ability to teach us the most. Be open to learning as you practice! We all do!

Right out of training, I had one of the most complex, time consuming and mind-boggling cases I've ever come across. It was a case of a child with an autosomal recessive condition where neither parent was a carrier. I was involved with long, agonizing conversations with the family about potential non-parentage, potential errors in testing, and to top it all, dealing with language barriers. In the end, it was a case of genetics that defied all the rules. Additional testing ultimately confirmed that the child's condition was the result of a new mutation followed by nondisjunction, resulting in UPD. That's one way to get published in your first year on the job!

I often tell students that the best thing they can do to continue to improve as genetic counselors is to self-assess after each case. Your training will prepare you for the profession but be open to learning through experience for the fine-tuning of skills over time.

Creating a Gender Inclusive Environment in Genetic Counseling

One of the most powerful and informative sessions during the 2022 annual PAGC conference was titled "Creating a Gender Inclusive Environment in Genetic Counseling". The session was moderated by Kelsey Bohnert (CHP) with speakers Andy Cantor (LabCorp Women's Health), Danielle McKenna (UPHS), and Elizabeth Sheehan (Magee-Women's Hospital). The PAGC Education Committee is working to make the recording of this session available to our membership.

As our understanding of sex and gender evolves, genetic counselors can approach their patient care with appropriate gender-neutral language. For individuals who identify as lesbian, gay, bisexual, transgender, or gender non-conforming, going to a healthcare appointment can be a daunting experience. At clinical practices that do not prioritize inclusive care, their experiences might be invalidated many times over—by the intake forms, by health history questions, and by the staff's failure to use gender neutral pronouns. Exclusive language can lead to individuals who are trans or gender non-conforming to avoid healthcare altogether.

The shift to *inclusive* language really doesn't cost us anything. And yet, it will earn genetic counselors so much currency in patient trust, which translates to better outcomes.

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The definitions below can help build a common vocabulary, and providers should always allow patients to describe themselves and should follow the patient's terminology. Mistakes will be made – apologize and move on. And when asking questions where sex assigned at birth is relevant to the genetic counselling session, be honest and transparent about your questions.

- **Sex Assigned at Birth; assigned female at birth; assigned male at birth:** The sex (male or female) assigned to a newborn based on their anatomy. May be referred to as birth sex, natal sex, biological sex or sex. Use *assigned female* and *assigned male* instead of biological female and biological male. In pedigrees, NSGC recommends noting someone as the gender they identify as (e.g., male with a square, female with a circle, and diamond for gender non-conforming), then adding either AFAB or AMAB under the symbol.
- **Cisgender:** An individual whose gender identity is the same as their sex assigned at birth. Use instead of "normal," "real" or "not trans."
- **Gender Identity:** An individual's personal sense of being a man, woman, both, neither, or another gender.
- **Gender Non-conforming (GNC):** An individual whose gender expression differs from society's perception or expectation for them as male or female. Gender expression can be different than gender identity. Gender identity is how a person views themself, whereas gender expression is how a person presents that gender and how society might view the person's gender. GNC individuals might identify as either gender, a gender different than male or female, or some combination.
- **Intersex:** An umbrella term for the variety of physical conditions in which the sexual or reproductive anatomy an individual is born with is not exclusively male or female. Many forms of intersex exist; it is a spectrum term, rather than a single category, and it is medical terminology that refers to anatomy rather than a term that defines gender identity or gender expression.
- **Nonbinary:** An individual who does not identify as a man or women.
- **Sexual orientation**: An individual who identifies as gay, lesbian, bisexual, or queer.
- **Transgender (abbrev. trans):** An individual whose gender identity differs from their sex assigned at birth, usually used when gender diverse traits are "persistent, consistent and insistent" over time. It is also used as a categorical term for gender identities other than male and female.

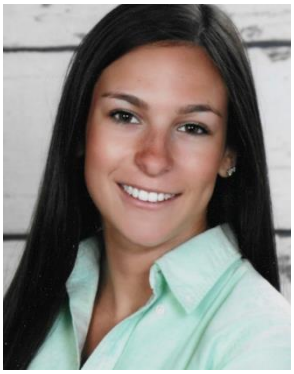
It is important to note that sex, gender identity, gender expression, and sexual orientation are all independent of one another. Assumptions cannot be made about other parts of an individual's identity based on information known about one category.

Some examples of inclusive language: *Genetic counselors can use these terms for all patients, not just with those individuals who "appear" to be gender diverse.*

- Parents, gestational or non-gestational parents – instead of mother or father
- Sibling(s) – instead of specifying brother and sister
- Spouse or Partner – instead of husband/wife
- Relationship status – instead of marital status
- "What sex were you assigned at birth?"
- "What is your current gender identity?"
- "What are your pronouns?" – instead of "What are your preferred pronouns?"

It is best practice to introduce yourself with your pronouns for every patient and give space for the patient to do the same, as certain situations may not be comfortable or safe to do so due to not everyone in the room being aware of someone's identity. It can also be a good idea to ask if there is a name the patient uses other than the name in the chart. Honesty, transparency, and most importantly, respect, build trust with patients.

Say this:	**Instead of saying this:**
Sex assigned at birth, AFAB, AMAB	"Real" sex, "real" gender, genital sex, biological sex, natal sex
Transgender person/Trans person	A transgender, a transgendered person, transsexual, tra**y
Transgender people/Trans people	Transgenders, transgendered people, transsexuals
Transgender man/Trans man	FTM, used to be a woman, born female, biological female
Transgender woman/Trans woman	MTF, used to be a man, born a male, biological male
Medical transition/gender affirmation treatment	Sex change, pre-operative, post-operative, "the surgery"



Featured Genetic Counseling Student: **Anna Platt**
University of Pennsylvania Genetic Counseling Program

What was your major as an undergraduate?
B.S. in Neuroscience from University of Delaware (2019)

What attracted you to pursue a career in genetic counseling?
As a long-time Special Olympics coach, I knew I wanted to help families with genetic conditions. After college, I worked as a Genetic Counselor Assistant for the Genetics team at Inova Fairfax Hospital. Although I had little knowledge about the genetic counseling field prior to becoming a GCA, the GCs and Medical Geneticists at Inova provided me with amazing mentorship and knowledge that allowed me to realize there was no other field I was meant to be in more than genetic counseling.

What field of genetic counseling are you most interested in post-graduation? My time at Inova allowed me to spend a lot of time observing NICU consults, as well as pediatric, neurology, and cardiology clinics. Although I've had the opportunity to see a variety of genetic counseling fields, I have fallen in love with both pediatrics and in-patient genetic counseling. I am attracted to the unique psychosocial and emotional impacts faced by families in the NICU and in-patient settings. I believe a genetic counselor's ability to ameliorate feelings of helplessness and frustration, and to empower parents to make informed decisions about their child's future care, can be the utmost rewarding experience. Additionally, I am drawn to the ability to maintain relationships with families and provide continuity of care in an outpatient pediatrics setting.

What has been the most valuable aspect of your training so far? One of the most valuable aspect of my training has been learning and hearing lectures from some of the best genetic counselors and doctors at two of the most renowned medical systems in the country, CHOP and Penn. I believe my clinical experiences at both of these institutions has been even more valuable as I combine my class knowledge with my developing genetic counseling skills.

Please provide a brief description of your thesis project. I am the 2021 Marie Barr Award recipient and will be completing my thesis with the supervision of Sarah Raible, MS, LCGC, Clinical Director of the Center for Cornelia de Lange Syndrome and Related Diagnoses. The Genomic Diagnostics in Cornelia de Lange Syndrome, Related Diagnoses, and Structural Birth Defects Study has just completed sequencing of 400 genomes from individuals around the world with a clinical diagnosis of Cornelia de Lange syndrome (CdLS), or a suspected CdLS-like condition. This historical project has been ongoing for over 20 years. By developing a research protocol to return these results and interviewing families about their experience and their perceived value of these results, we hope to demonstrate the impact of this multidecade-long study on the research community and CdLS families.

PAGC Committees

Volunteers are always welcome!

Contact committee chair if you are interested in being involved

Education

Chairs:
Amanda Back (amanda.back@jefferson.edu)
Dana Farengo Clark (dana.farengoclark@pennmedicine.upenn.edu)

- Plan annual PAGC meeting
- Plan CEU-eligible webinars throughout the year

Professional Issues

Chairs:
Becky Belles (becpitt30@gmail.com) and
Livija Medne (medne@chop.edu)

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

Genetic Services

Chair: Kallyn Stumm (kallyn.stumm@gmail.com)

- Implemented Professional Status Survey for PA GCs and analyzed results (see PAGC website)
- Continue to evaluate GC services in PA

Membership

Chair: Susan Walther (susanwalther1203@gmail.com)

- Implemented new website design and maintain content
- Manage e-blast communications
- Create PAGC newsletter

Diversity, Equity & Inclusion Working Group (currently, an ad hoc committee)

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

- Develop framework for initiatives to improve DEI in the PA GC community
- Diversify “best practices” for GCs as they relate to minority patient populations
- Develop education on DEI issues