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Official Newsletter of the Pennsylvania Association of Genetic Counselors

PAGC News

PAGC
Pennsylvania Association of Genetic Counselors

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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2021 Arch Street
Philadelphia, PA 19103

PAGC Website:
www.pennsylvaniagc.org

PAGC held the 9th Annual Conference April 3-4, 2025 Philadelphia Marriott West Conshohocken, PA

What a great experience it was to be able to connect with colleagues for the annual conference! The PAGC Education Committee met the challenge of providing quality presentations for in-person and virtual attendees. A total of 220 people registered this year; 95 genetic counselors obtained CEUs. All around – a successful, engaging event. We look forward to next year!

Continue reading for topic highlights from the conference.

Thank you to our 2025 Sponsors!

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Summaries of Presentations from the 2025 PAGC Annual Conference

96041 Genetic Counselor CPT Code Implementation

***Speakers*:**
Livija Medne, MS, CGC (Children's Hospital of Philadelphia)
Rachael Brandt, PhD, MS, CGC (Main Line Health)
Megan Betts, MS, CGC (WellSpan Health)
Seth Lascurain, MS, , MPH, CGC (UPMC MFM Harrisburg)

Summarized by Cassy Gulden, MS, CGC

The new 96041 CPT code for genetic counseling went live in January 2025. There are a couple of important differences between the old 96040 and the new 96041 code. The new code is meant to be used by *trained* genetic counselors. It should not be used by other professionals who perform genetic counseling. Another difference between 96040 and 96041 is that the new billing code now includes preparation time, face-to-face time, and post-visit time/documentation on the same day as the patient's visit. Similarities between the codes include: the number of units billed is still in 30-minute increments and phone/video visits can be billed.

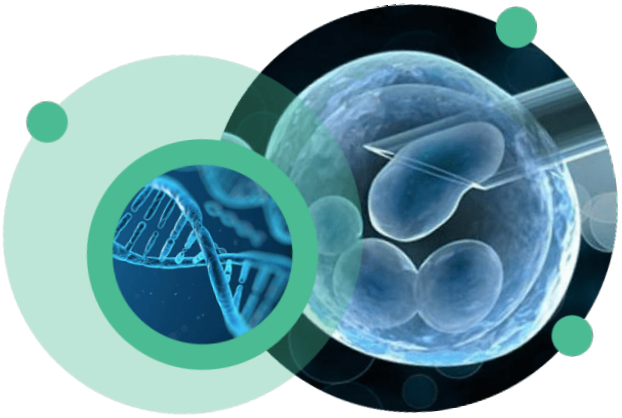
Panelists discussed different impacts on implementing this code in their respective clinics. Several were able to provide preliminary data showing an increase in billing units in the first quarter of 2025 as compared to 2024. Some clinics are now able to bill for telemedicine results or follow-ups that they may not have been able to bill with the old code. Genetic counselors need to find appropriate ways to track their time and document it in their patient notes. One thing to keep in mind is how the policies for the new CPT code may affect our patients with Medicare and Medicaid. While it is important to be recognized for all the work we do for our patients, billing some patients for more time may result in burdening them with higher out of pocket costs. Again, this all circles back to our efforts for CMS recognition. Overall, the new CPT code 96041 is a major step forward for genetic counselors gaining appropriate recognition for our time and efforts to provide quality patient care.

MEMBERSHIP NEWS

PAGC continues to increase our membership. 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.

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/

Updates in Fertility Genetic Testing



Speaker: **Alex Morgan, MS, CGC**
(Geisinger Health System)

Summarized by Rebecca Oberschmidt, MS, CGC

There are many indications for an individual or couple to seek fertility genetic counseling, including intent on using donor egg or sperm, consideration of implanting a low mosaic embryo, recessive carrier couples, an individual with a genetic condition, and a balanced translocation carrier. There are also several forms of preimplantation genetic testing, including PGT-A, PGT-M, PGT-SR, PGT-PRS (polygenic risk score), and PGT-HLA.

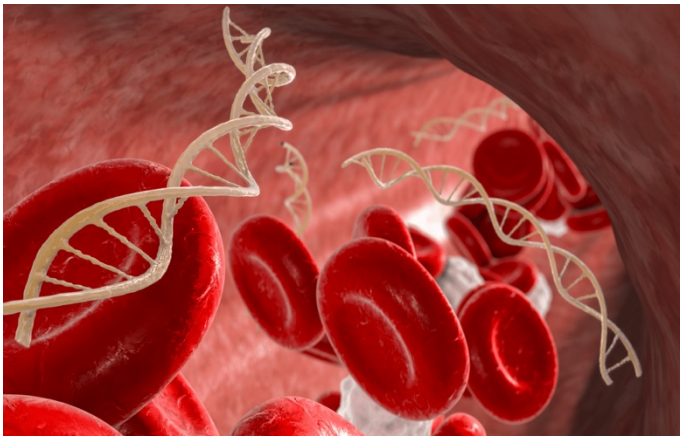
The goal of **PGT-A (aneuploidy)** is to reduce miscarriages and increase live births. The results are reported as euploid, aneuploid, or mosaic, where mosaic can be low-level mosaic (20-40/50%) or high-level mosaic (40/50-80%). It is important to remember that PGT-A is still a screening test and; therefore, the results may or may not accurately reflect the genetic status of the embryo. There is emerging technology of noninvasive PGT-A, where an embryo is rinsed, and media collected from that rinse and analyzed for cfDNA. This process would decrease cost and risks to the embryo.

PGT-SR (structural rearrangement) is used to detect unbalanced translocations and can help to explain lack of blastocyst progression. However, it cannot distinguish balanced translocation carriers from other euploid embryos.

PGT-M (monogenic condition) is used when there is a known variant in the parent(s). The process of PGT-M requires lab approval in each case. This process also requires parental samples so that the lab is equipped to identify the variant(s).

PGT-HLA (human leukocyte antigen) is used often when a sibling needs a bone marrow or cord blood transplant, which is often completed alongside PGT-M for a known genetic condition. The HLA region of biopsied cells is analyzed.

There are conflicting studies on mosaic embryos regarding live birth and miscarriage rates, and it is unclear what these results mean and what they can look like in practice. There are concerns for risks of UPD, confined placental mosaicism, and fetal growth restriction. Some clinics do not allow for transfer of mosaic embryos, and there is limited data for patient considerations when transferring mosaic embryos. Additionally, segmental mosaic embryos are more likely to be a false positive. Genetic counseling after mosaic embryo transfer should include a recommendation that amniocentesis is the preferred sample type, along with microarray, FISH, and UPD studies.



Updates in cfDNA for Prenatal Genetics

Speakers:
Madalyn Charnego, MS, MPH, CGC
Rebecca Oberschmidt, MS, CGC
Seth Lascurain, MS, MPH, CGC
(UPMC MFM Harrisburg)

Summarized by Madalyn Charnego, MS, MPH CGC

Prenatal cell-free DNA (cfDNA) Screening has traditionally been used as a method of screening pregnancies for the risk for common aneuploidies. In recent years, it has also evolved to include single-gene cell-free DNA (**sgcfDNA**) screening for fetal conditions such as alloimmunization and recessive conditions. Prenatal sgcfDNA analysis can be completed through various methods, including dosage analysis and variant detection. Both methods have been used by the labs developing this technology, and both methods have limitations. Currently, there is no industry standard for this technology.

In a clinical setting, prenatal sgcfDNA can be used for a variety of indications including personal or family medical history, specific ultrasound findings, and Rh incompatibility. Additionally, prenatal sgcfDNA can be a shorter pathway to learning more about the risk for a pregnancy when a partner is not available. For situations of alloimmunization, there has been a proposal to use low risk results to reduce the use of RhoGAM and potentially save this limited resource. According to ACOG, the use of sgcfDNA technology to prioritize and conserve RhoGAM should be taken into consideration.

As illustrated by a variety of case examples, prenatal sgcfDNA is a useful methodology for screening a pregnancy. Highlighted by one case, it is important to remember that this methodology is still a screening and not diagnostic for any condition. The case involved a patient with high risk prenatal sgcfDNA results for sickle cell disease in the context of a partner who is not a known carrier for sickle cell trait. The patient completed diagnostic testing for the pregnancy, which identified one of the first false positive results from the sgcfDNA methodology that the lab used.

In summary, prenatal sgcfDNA is an emerging technology with complex methodologies. These methodologies for analysis have technical barriers and given this, an industry standard has not been set for these screening methods. Prenatal sgcfDNA has a variety of uses including screening for conditions in a patient's personal or family history, as well as evaluating Rh incompatibility. Finally, as with all screenings, there is a chance for false positives or negatives, and follow-up genetic counseling should be provided for patients to discuss any potential recommendations for confirmatory testing.

Delivering Healthcare to the Plain Community

***Speakers*:**
Susan Walther, MS, CGC (Clinic for Special Children)
Amy Albright, MS, CGC (Clinic for Special Children)
Cate Walsh Vockley, MS, CGC (UPMC, Children's Hospital)
Ruth Zook, OOA (Old Order Amish)

Summarized by Susan Walther, MS, CGC

Anabaptism is a Christian movement that emerged from the Protestant Reformation in the 16th century. Anabaptists believe in adult baptism, as opposed to infant baptism, when an individual can confirm their commitment to the church and the teachings of the Bible. Additionally, Anabaptists believe in living a simple, humble life, believe in minimal interface by government in religious practice, and believe in nonviolence. Key Anabaptists groups include Mennonites, Amish, and Hutterites. Throughout Pennsylvania, the largest Anabaptist communities are Mennonite and Amish.

Anabaptists are more commonly described as "Plain" due to their simple style of dress of muted colors and black skirts and trousers. Mennonite women wear hair coverings of different styles and are more often seen wearing dresses with prints. Amish women wear heart-shaped bonnets that cover most of their hair and always wear solid color blouses under black pinafore dresses. Mennonite men wear black felt hats, and Amish men wear straw brimmed hats; both conservative Mennonite and Amish men wear suspenders instead of belts.

The Clinic for Special Children in Gordonville, Lancaster County, and the Plain Community Translational Medicine Program at UPMC strive to serve Plain families with children diagnosed with genetic conditions by delivering healthcare that is attuned to their cultural beliefs. There is a strong collaborative approach between the Plain healthcare entities in Pennsylvania, and the collaboration extends throughout the United States to include nine organizations in the Plain Community Health Consortium. The collaborative approach has enriched understanding of genes and variants that are most prevalent in the Plain community with distinct genetic differences between Amish and Mennonite groups. These genetic differences are due to bottleneck effect of persecuted Anabaptist populations leaving Europe and emigrating to the United States in the 16th century, followed by founder effect as groups of individuals formed communities. The Clinic for Special Children developed a test specific to Plain genetics called "Plain Insight Panel™"; this test is designed for carrier screening and for diagnostic purposes.

It is becoming more likely that genetic counselors will interact with individuals and families from the Plain community due to the growth of settlements throughout Pennsylvania. And it is important to be aware of Mennonite and Amish ancestry and the genetic specificity as some young adults are choosing to live more modern lifestyles outside of the cultural norms of their childhood.

Helpful tips on next page

Helpful tips for delivering healthcare to the Plain community

- Parents often prefer to only do what is necessary for medical intervention
- Reserved, inward reactions; silence often represents disagreement or lack of understanding – ask “What questions might you have?” rather than “Do you have any questions?”
- Grief is quiet; acceptance of God’s plan
- Families have tremendous support from their community. Don’t assume families won’t follow up or be able to accommodate medical equipment in their home
- Families appreciate phone calls if a clinic visit is not critical; families have to pay to hire a driver (Amish) and arrange childcare
- Provide information in writing whenever possible; parents appreciate reading to learn new information, and they like to be able to share information with community members; young couples often look to their parents for advice
- Plain families are self-pay; signing the 4029 form makes adults ineligible for Medicaid and Medicare; cost is a consideration when deciding about medical services

Common last names of Plain families

Amish	Mennonite
Beiler/Byler	Brubaker/Brubacker
Esh/Esch	Burkholder
Fisher	Hoover
Glick	Horning
Hostetler	Horst
Kauffman	Hurst
King	Leid
Lapp	Martin
Miller	Newswanger
Riehl	Nolt
Smucker	Reiff
Stoltzfus/Stoltzfoos	Sensenig
Troyer	Shirk
Yoder	Weaver
Zook	Zimmerman

The Ehlers-Danlos Society App

Jessica Goehringer, MS, CGC

I had the pleasure of working with the Ehlers-Danlos Society on the creation of an App that is designed to **“unite and support individuals living with the Ehlers-Danlos syndromes (EDS) or hypermobility spectrum disorders (HSD) worldwide”.** Empowering the EDS community has been a passion of mine, as has mentoring. I combined those two interests and worked with two genetic counseling graduate students on a thesis project that assessed the quality and utility of a prototype of a diagnostic tool I designed that is intended to support diagnosis of hypermobile EDS. I shared the results of the project with the EDS Society, and our synergistic goals culminated in working together to release a patient- and provider-facing App at the end of 2024. The App includes patient and provider resources, sections that allow users to document and export their personal health profile (health history, emergency care instructions and contacts, allergies, conditions, past surgeries, etc.), symptoms, and medications, as well as set medication reminders. There are diagnostic tools sections that house diagnostic checklists for EDS/HSD. The App is envisioned to expand in resources to include the ability to connect globally with individuals who share similar EDS diagnoses.

To download the App: The Ehlers-Danlos Society App - The Ehlers Danlos Society

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 (terks1@chop.edu)

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

Professional Issues

Chair: Becky Belles (rsbelles@geisinger.edu))

- Work to update GC licensure in PA
- Examine barriers to credentialing of GCs in PA
- 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)

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

Justice, Equity, Diversity and Inclusion

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

Please visit the new dedicated website page for J.E.D.I information:
https://www.pennsylvaniagc.org/jedi