

UPDATES

UPenn MSGC Program



Our First Five Years at Penn

KATHLEEN VALVERDE, PHD, LCGC



As we celebrate the fifth graduating class of the University of Pennsylvania's Master of Science in Genetic Counseling Program, I want to reflect on how far we have come and where we are headed.

This newsletter highlights our activities and accomplishments over the past five years. In just a few years, our program has grown stronger and become more deeply enriched by the support of the vibrant genetics community at UPenn and CHOP. This enduring commitment has been instrumental in building a program that stands out in excellence and impact. We have expanded our curriculum, strengthened our partnerships, and proudly watched our alumni step into leadership roles within the genetic counseling community and the program.

We pause at this milestone to recognize and thank the many individuals who make this program possible. To our dedicated instructors, staff, clinical supervisors, research and professional mentors, committee members, and supporters, your tireless efforts and belief in the program and students have shaped every part of the journey over the past seven years. Your unwavering dedication to excellence has left an enduring imprint on every graduate.

At this year's graduation, I shared with our newest alumni the analogy that genetic counseling is like a kaleidoscope. Each patient brings a unique pattern, a mosaic of genetics, culture, lived experiences and emotions. The genetic counselor's role is to help bring clarity to that pattern and find meaning in what may at first appear as scattered fragments. We teach our students that a single variant can mean different things depending on the context. The same information, viewed through various lenses, may have a different interpretation. Sometimes the most meaningful thing we can offer is not certainty but clarity, compassion, and the courage to sit with uncertainty.

To our graduates, thank you for trusting us with your education and growth. You prove what is possible when passion meets opportunity and hard work. You are the ambassadors of this program, and your contributions to patient care and the profession continually affirm our vision and values.

As we look ahead, we remain inspired by the experiences of our first five graduating classes. Their commitment and achievements continue to guide and shape our future. Today, we celebrate our 87 UPenn alumni and almost 400 total alumni, everything we have accomplished together, and the growing legacy that strengthens with each graduating class. I am confident that the best is yet to come!

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Introducing Assistant Program Director Tiffiney Hartman



Tiff Hartman, PhD, LCGC, was hired as the Associate Program Director for the Career Ladder Education for Genetic Counselors (CLEP-GC) grant in January 2023, and, upon Laura Conway's retirement in June 2024, she became Assistant Director for the MSGC program. Tiff took over teaching Laura's Advanced Genetics and

Genomics course and works closely with Rebecca Mueller on the Research Methodologies course, in addition to being an MSGC thesis advisor for multiple students and continuing her CLEP-GC duties, which include directing the ART-GC certificate program and the CEU series. (More on this later.)

Tiff started her graduate studies with a broad interest in disease at the molecular biology level and devoted her career to studying human genetic conditions, cancer, and stem cell research, and educating others on these topics. After receiving her PhD in Molecular, Cellular & Develop-mental Biology, she worked as a postdoctoral fellow and Assistant Research Professor for several years, investigating the molecular and signaling mechanisms in human genetic diseases as a basic scientist. Tiff studied a variety of conditions on the bench over the years, including Tuberous Sclerosis Complex, Polycystic Kidney Disease, and pancreatic cancer utilizing cell culture, Drosophila, and mouse models.

In 2017, Tiff returned to school to obtain an MS degree in Genetic Counseling at Arcadia University, with the goal of continuing her passion for research on and education about human genetic conditions while helping individuals understand and cope with their diagnoses. After graduation, Tiff joined the Roberts Individualized Medical Genetics Center at CHOP, where she worked both in the clinic as a GC and as part of the research team before taking on her roles with the Penn program. Tiff does not anticipate any more career pivots as she feels most at home teaching and working with students on their thesis projects.

Transitions



Katherine Nathanson, MD

Dr. Nathanson became chair of the program's Advisory Committee in 2022. Dr. Nathanson is the Pearl Basser Professor for BRCA-Related Research and Deputy Director of the Abramson Cancer Center, among many other titles, and an internationally recognized expert in cancer genetics and genomics. Her guidance and leadership is greatly appreciated in the MSGC program. In addition to her role on the committee, Dr. Nathanson is an enthusiastic supporter of GC student research and clinical training at the Basser Center.



Juanita Neira, MD

We welcomed Dr. Neira as the program's new Medical Director this year. She is a Clinical and Biochemical Geneticist who joined CHOP as an attending physician with the Division of Human Genetics. She is an Associate Professor of Clinical Pediatrics at the Perelman School of Medicine. Previously at Emory University School of Medicine, Dr. Neira has a focus on inborn errors of metabolism.



Ian Krantz, MD

During more than 25 years at UPenn and CHOP, Dr. Krantz championed genetic counselors and the MSGC program. He served as the program's medical director for over a decade and was instrumental in bringing the program from Arcadia to Penn. He is now the Division Chief of Pediatric Genetics and Genomics at Northwell Cohen Children's Medical Center, the System VP for Pediatric Genetics, and professor at the Zucker School of Medicine at Hofstra/Northwell



Laura Conway, PhD, LCGC

Laura retired after 20 years (2004-2024) in program leadership. Colleagues and alumni from across the years came to thank her and wish her the very best at a farewell event in May 2024. Her contributions are evident in every part of the program, and, most important to her, with all the students she taught and mentored.

The Long View



Daniel Rader, MD

In 2018, Dan Rader, MD, had a vision to integrate genetic counseling education across the University of Pennsylvania and Penn Medicine, while building a more diverse and professionally supported workforce. Under his leadership, the MSGC program was launched at the Perelman School of Medicine in 2019, laying the foundation for Penn to become a national leader in the field. This vision was realized by recruiting Kathleen Valverde, PhD, LCGC, as the inaugural program director, and the other key members of the Arcadia University program: Laura Conway, PhD, LCGC; Lisa Kessler, MS, LCGC; and Jo MacKenzie.

With the generous support of the Warren Alpert Foundation, Dan Rader and Kathleen Valverde, as co-principal investigators, secured two major grants: The Alliance for Genetic Counseling Fellowship, launched in 2021 with \$9.5 million in funding, and the Career Ladder Education Program for Genetic Counselors, awarded in 2022 with \$9.7 million. The Alliance grant provides 40 genetic counseling students with a scholarship and an educational fellowship, and the Career Ladder supports the participation of 50 genetic counselors in the Advanced Research Training Certificate Program, with protected time to develop an independent research project. These initiatives have been transformative and have accelerated the MSGC program's impact on the genetic counseling field by enhancing education, expanding access, and fostering innovation.

We also thank Dr. Rader for his support in securing standing faculty academic appointments for Kathleen Valverde and Rebecca Mueller, PhD, LCGC, and adjunct faculty appointments for Tiff Hartman, PhD, LCGC, and Lisa Kessler, MS, LCGC, in the Department of Medicine.

Supporting Belonging: Establishing Mentoring Relationships in Genetic Counseling

In 2020, shortly after the program moved to UPenn, and seemingly overnight, COVID upended many of our familiar social and educational practices. Even after we all became Zoom experts, connections remained challenging. So, we began a mentoring initiative with the goal of connecting incoming genetic counseling students with practicing genetic counselors who could support them as they entered the MSGC program.

Now five years on, this initiative has grown into a formal mentorship program with established guidelines, and it has expanded to the other four GC master's programs in the Alliance for Genetic Counseling Fellowship. Lisa Kessler, MS, LCGC, Associate Director of the UPenn MSGC program, has been the driving force behind this development. As the mentoring program became more established, she wanted to learn more about the scholarship of mentoring, and she is now an Education and Leadership PhD candidate at Pacific University,

My research aims to better support genetic counseling students by exploring of sense of belonging in their master of genetic counseling programs and to develop more effective mentorship in genetic counseling graduate education through exploration of mentee and mentor experiences of being in mentoring relationships."

- Lisa Kessler



Mentee Shannon Gray, '23, now at GC at Penn Medicine, with Mentor Meron Azage, MS, LCGC, from the Penn Department of Neurology

Lisa is also interested in learning about the meaning that genetic counselors make from working with students and the impact of these relationships on the mentor as well as on the mentee. And, she is curious about the differences between supervisory relationships and research mentoring relationships and how these are distinct from long-term mentoring relationships where students work with the mentor for one-to-two years. Stay tuned for more on her work. Lisa expects to complete her PhD in 2026.

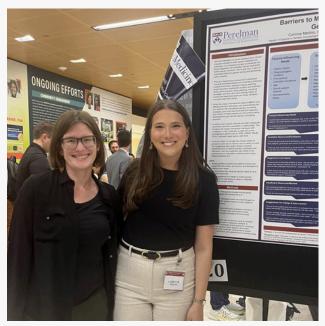
Rebecca Mueller Receives Prestigious NIH Award

MSGC Program Director of Research, Rebecca (Becca) Mueller, PhD, MSGC, recently received a K01 Career Development Award from the National Human Genome Research Institute for her work entitled "Childhood-onset genetic conditions: Planning for the future amid therapeutic innovation."

In a period when the outlook for many childhood-onset genetic conditions is changing with the introduction of disease-modifying therapies, Becca's work examines how affected adults understand the shifting prognostic implications of their diagnoses and make future-oriented life decisions. She aims to identify novel clinical opportunities to support patients as they navigate life plans amid prognostic uncertainty. Her K01 also provides support for career development in qualitative methods and disability studies.

Disability studies has been a focus of her graduate work at the University of Pennsylvania, where she earned a PhD in the History and Sociology of Science in 2021 and completed a postdoctoral fellowship in Ethical, Legal, and Social Implications of Genetics and Genomics.

Becca graduated from the Arcadia University genetic counseling program in 2011 and then worked clinically Penn's Basser Center for BRCA, before beginning her doctoral training. She has been involved with the MSGC program since 2015 as an instructor and thesis advisor and joined the Penn MSGC leadership in July 2024. In July 2025, she was appointed Clinician-Educator Assistant Professor in the Department of Medicine - Translational Medicine and Human Genetics. She will continue as Research Director for the MSGC program and the ART-GC certificate program and has already made great progress in developing thesis goals and guidelines to support the students in bringing their research to publication.



Rebecca Mueller, PhD, MBE, LCGC, with Corinne Merlino, Class of 2025, at Penn Medicine Dept of Medicine Research Day

especially important role in rare disease, and I am eager to bring advanced qualitative methods like disease concept models into the MSGC thesis curriculum. I also hope to expand offerings in Penn's Career Ladder Education Program for Genetic Counselors (CLEP-GC) to help equip GCs to work in roles focused on the development and implementation of targeted therapeutics."

- Rebecca Mueller

A Statement of Values is a Path for the Future



Associate Program Director Lisa Kessler, MS, LCGC, has been an active member of the Diversity, Equity, Inclusion and Justice Committee for the Genetic Counseling Educator Association for the past several years. The GCEA, previously the AGCPD, is a collaborative group of graduate-level educators involved in the 60+ accredited genetic counseling master's programs in the US and Canada. She and another member of the committee, Riyana Babul-Hirijand, Assistant Program

Director of Operations for the M.Sc. Program in Genetic Counselling at the University of Toronto, were honored to be tapped to create the organization's value statement, which Lisa calls "one of my proudest accomplishments on this committee." The statement they developed was reviewed and edited by the rest of the members of the DEIJ Committee, approved by the Board of Directors, and put into practice this year. You can read the full Statement of Values at https://educategc.org/statement-of-values/.

Beyond Classes and Clinic: A Student's Perspective

ISABELLA BROWN, UPENN MSGC '26



Isabella Brown (right) at the Penn Community Health Fair with Genetic Counselor Jessica Long, MS. LCGC

Learning about genetic counseling in classes is one thing, but applying those skills always refreshes me with meaning and purpose. Internships are certainly one way to achieve that; however, I am finding more and more that outreach is extremely rewarding in a whole other way. When we see patients, we are seeing those who had the right referrals or influences in their lives to get them to an appointment with us. By running an outreach table for breast cancer screening and genetics awareness, though, the Basser Center for BRCA allows us to speak with people who may never enter our office otherwise. It's a safe space for them to talk with professionals about their risks, without cost or obligations. Additionally, the table has specific resources for the most underserved populations in breast cancer - Black and Latina women - so we can communicate in a relevant way to them.

Every time I work at this table at outreach events, such as the Penn Community Health Fair a few weeks ago, I am thrilled to speak with people with many different concerns, those who are worried about their family history of cancer, but didn't know who to ask about it; those who had testing years ago and have been wondering about updated testing; those who never knew their risk was elevated by their family history; and even those who only hesitate due to their understanding of cost. It's incredibly fulfilling to feel we are helping people who want this knowledge and may not ever have received it without our outreach.

I think this work is exactly what the Basser scholarship is about. By expanding the pipeline of genetic counselors, the Basser Center is helping to reach all people and empower them with critical health information. The Basser Center is breaking down barriers and increasing access to genetic counseling and testing to help students become genetic counselors who will continue this work. I take this responsibility seriously and hope to continue reaching out to the groups who need it most, to make care more accessible and less stressful.

Isabella Brown is the Class of 2026 recipient of the Basser Center Genetic Counseling Scholarship. Generously funded by members of the Basser Center for BRCA's Advisory Board, the scholarship covers the cost of tuition for both years of the MSGC program at UPenn.

Basser Center for BRCA Genetic Counseling Scholarship



A new genetic scholarship envisioned by Susan M. Domchek, MD, the Executive Director, and funded by members of the Advisory Board of Penn Medicine's Basser Center for BRCA—a comprehensive center for the research, treatment, and prevention of BRCA-related cancers—is set to make a difference in this vital field of preventative medicine. First awarded in 2022, the endowed scholarship is designated for individuals representing various perspectives and experiences who are pursuing a master's degree in genetic counseling at the Perelman School of Medicine. The scholarship covers full tuition costs for the two-year graduate program. This vital aid will create opportunities to increase diversity in the genetic counseling workforce by offsetting costs that, for many promising scholars, represent an insurmountable obstacle to entering careers in this field.

Building Community Across MSGC Programs

Through the Alliance for Genetic Counseling Fellowship

The Alliance for Genetic Counseling Fellowship (AGCF) is funded by a generous grant from the Warren Alpert Foundation to the University of Pennsylvania MSGC program and Kathy Valverde, Pl. It is a consortium of five accredited genetic counseling master's programs (Boston University, Sarah Lawrence College, Rutgers University, the University of Maryland, and the University of Pennsylvania) with a goal to recruit and graduate students from backgrounds underrepresented in the field. Creating community among students who live in different locations and attend different programs so they can learn from and support each other, not only as students, but ultimately as professionals too, is essential to its success.

The students gather in person twice each year for retreats that aim to promote a sense of community, establish a common language to assist them in having supportive and difficult conversations, provide space for them to listen and learn from one another, and create a community of practice by articulating shared concerns and values. In addition, virtual monthly meetings allow the first-year cohort of 10 students among the five Alliance programs to participate in workshops and develop a community of peers. We asked UPenn Site Coordinator, Meron Azage, MS, LCGC, and Thomas Washington, a Warren Alpert Scholar and student in the UPenn MSGC program's Class of 2026, to share their thoughts on community building.

MERON AZAGE, MS, LCGC

Since October 2023, I have facilitated a monthly virtual fellowship meeting for our Warren Alpert scholars with the goal of creating a supportive space that fosters community building and connection. These gatherings encourage scholars to bring their authentic selves. We openly discuss challenging and controversial topics with radical honesty while adhering to norms of confidentiality and mutual respect established from the outset. A scholar once shared with me that these meetings helped ease her anxiety about attending our first in-person retreat, as she had established connections with the other scholars. Through open dialogue on the successes and challenges of navigating graduate school, scholars develop resilience and strengthen their ability to thrive in their future careers as genetic counselors.

Meron Azage is the Associate Director of Genetic Counseling in the Department of Neurology in the Perelman School of Medicine. She joined the staff of the AGCF in 2023 as the site coordinator for the University of Pennsylvania.

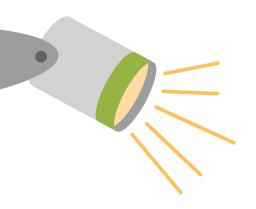


THOMAS WASHINGTON, UPENN MSGC '26

As first-year Warren Alpert Scholars, we met once a month in fellowship meetings. These meetings were a place where we could discuss life as genetic counseling students and talk about current events. The meetings fostered connection between me and the other scholars as they forced us to take a short break from our busy schedules and come together to talk. Meron Azage, a genetic counselor in the Department of Neurology in UPenn's Perelman School of Medicine, led our monthly meetings and always facilitated conversation with an icebreaker question or topic. However, over time, the conversations in these monthly meetings seemed to flow more naturally on their own as scholars got to know each other. I feel that as a group of first-year WAF scholars, we formed our own community. This community provides another support system for students as they go through a challenging academic program. Importantly, not only do members of this community have commonality in their academic and professional pursuits but also in the fact that they come from varied backgrounds. Therefore, the WAF scholar meetings, and resulting community, provide the opportunity for commiserating about shared experiences.

Thomas Washington is a member of the Class of 2026 in the University of Pennsylvania MSGC program and a recipient of an AGCF Fellowship.

Warren Alpert Scholars and program staff gather at the fall 2024 retreat in Philadelphia.



Spotlight Q & A: A Model for Mentoring GC Student Research

When the MSGC program moved to UPenn in 2019, program leaders shared a vision with those at Penn that our students would become researchers and scholars, integrated into many departments and fields of inquiry. Nowhere has that happened more smoothly than with **Bryson Katona**, **MD**, **PhD**, and the team of genetic counselors who work with him on research projects in gastrointestinal cancer: **Jessica Long**, **MS**, **LCGC**; **Jessica Ebrahimzadeh**, **MS**, **LCGC**; **Jackie Cappadocia**, **MS**, **LCGC**; and **Derek Mann**, **MS**, **LCGC**.

Dr. Katona is the Jeffery and Cynthia King Associate Professor of Lynch Syndrome Research and an Associate Professor of Medicine and Genetics at the University of Pennsylvania Perelman School of Medicine, where he serves as the Director of the Gastrointestinal Cancer Genetics Program, Director of the Gastrointestinal Cancer Risk Evaluation Program, and Director of Penn Medicine's King Lynch Syndrome Program. He is the current president of the Collaborative Group of the Americas on Inherited Gastrointestinal Cancer (GCA-IGC). And with all that, or because of all that, he is exceptionally supportive of genetic counselor researchers and GC student researchers.

This team has mentored an MSGC student research project every single year since our move to Penn, working with Jeshua DeJesse, '21, Samantha Williams, '22, Isabel Anez Bruzual, '23, Sam Levy, '24, Grace Snyder, '25, and this year both Lauren Cuff, '26, and Samantha Kasselman, 26, which is exceptional. They have been challenging and interesting projects with excellent presentation and publication outcomes, exactly what we were hoping for when we moved to Penn.

So, we wondered, how do they do it?

- How do they all work together, teach and mentor the students so that they make a real contribution, "right-size" the projects for completion in a year, and see them through to publication?
- What wisdom and experience can they share with other researchers who are potential mentors?

We asked, and they told us. Turn to page 16 for the full interview.



Student thesis research publications: <u>Jeshua DeJesse</u>, <u>Isabel Anez-Bruzual</u> & <u>Samantha Williams</u>. Click on the name or image to see the full paper.

Continuing Education for Genetic Counselors

TIFF HARTMAN, PHD, LCGC

In 2023, Kathleen Valverde, PhD, LCGC, Dan Rader, MD, and the UPenn MSGC Program were awarded a grant from the Warren Alpert Foundation to create the Career Ladder Educational Program for Genetic Counselors (CLEP-GC). They formed a consortium of expertise with four genetic counseling master's programs located at Baylor College of Medicine, Northwestern University, Vanderbilt University, and the University of Washington.

One arm of the grant supports the development of five ten-hourlong continuing education courses for genetic counselors. We have leveraged the expertise and experience of the principal investigators and program directors from the consortium sites to create this high quality impactful educational series. The courses are free for anyone to take, available to GCs anywhere in the world, and approved by the NSGC for 1.0 CEU credit. NSGC CEU fees are paid in full by the grant for the first 250 individuals who complete each course.

The first course <u>Bridging the Gap: A Clinical Genetic</u>
<u>Counselor's Guide to Molecular Diagnostics and Variant</u>
<u>Interpretation</u> was coordinated by Sara Reichert, MS, LCGC, a genetic counselor with the Division of Genomic Diagnostics at CHOP and made available in March 2024. (More than 250 people have completed this course, so viewers can still watch the course for free, but would pay the NSGC \$35 for their CEUs). Feedback has been overwhelmingly positive.



Based on evaluations completed by the first 251 participants, 93.6% rated the course extremely or very useful and 98.8% agreed that it enhanced their professional expertise." - Tiff Hartman



The second CEU course <u>Genes, Choices, and Ethics:</u>
<u>Navigating the Moral Dilemmas in Genetic Counseling</u> opened in May 2025. Feedback has again been incredibly positive. Both courses are available by clicking the titles or using the QR codes on the flyers.

The grant funds five total courses, and the next two are currently in the design and record process. *Genetic Counselors as Educators: Practical Teaching Strategies for Genetic Counselors* is on track for fall 2025, and *Genetic Counselors and Use of BIG Genomics Data Sets* for spring 2026.

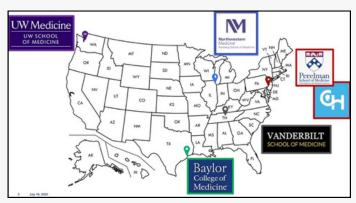
We'd love to hear topic ideas from our GC alumni and community for the fifth and final series – stay tuned!

Advanced Research Training for Genetic Counselors

TIFF HARTMAN, PHD, LCGC

A second arm of the CLEP-GC grant from the Warren Alpert Foundation supports an online certificate program for practicing genetic counselors called the Advanced Research Training for Genetic Counselors program (or ART-GC). Practicing GCs from the five consortium sites can apply for the one-year program, and two from each site are selected each year over the five years of the grant. Impressively, the grant covers the creation of the online courses, full tuition for all participants, 50% full time effort for one year to provide for protected research time, travel funds for conferences, and \$5,000 each in research funds. Participants are also matched with a career mentor for the course of the program.

Each participant takes three required courses: Research Methodologies (course director Tiff Hartman), Ethical Legal and Social Implications of Genetics (course director Rebecca Mueller), and Grant and Manuscript Writing (course director Tiff Hartman). They select an elective course from an array of options available online through PSOM's Master's and Certificate programs. Two electives were created for ART-GC: Biomedical Informatics for Genetic Counselors (course director Marylyn Ritchie), and, coming in spring 2026, Mixed Methods Research for Genetic Counselors (course director Shimrit Kedem). Additional online electives are available through Health Care Innovation, Translational Research, Public Health, and Regulatory Affairs, and cover topics such as implementation science and clinical trial management. Participants take the primarily asynchronous online courses, design and work on independent research projects, and meet each week over Zoom as a group to discuss course content and research progress and roadblocks.



Location of consortium sites.



Director Tiff Hartman, second from right, with ART-GC students at the 2025 American College of Medical Genetics Meeting in Los Angeles, CA,

The first two cohorts have completed the program and appreciate their new research skills and the networking connections they formed with each other. Participants report having gained confidence to pursue and lead their own research projects and feeling validated as researchers and empowered to submit their work to high powered journals. This newfound scientific self-assurance is reflected in some of the ART-GC outcomes we have been tracking, which include seven published manuscripts with several more in preparation, many conference abstracts, guest lectures, job promotions, PI status for IRB applications at institutions that typically do not give this status to those without a terminal advanced degree such as PhD or MD, and grant applications and awards. There is a lot going on, and it's only been two years!

Congratulations to ART-GC graduates from CHOP/Penn: Alyssa Rippert and Anna Raper from year one, and Natalie Burrill, Elizabeth McCormick, and Sarah Ruggiero from year two. Year three participants, Avi Anantharajah and Jamie Peterson, begin the program this August.

Learn more about the ART-GC program at https://clepgc.med.upenn.edu/certificates.

GC Students Cross Paths with a

Much-Admired Researcher

Wendy K. Chung, MD, PhD, came to Penn this spring for the Distinguished Speaker Series Genetics Seminar. Students in the MSGC program were thrilled to attend her lecture "Pediatric Genomic Medicine: N of 1 to population health." It wasn't just that they respected her status in the field, currently as Chief of the Department of Pediatrics and a named professor at Harvard Medical School. Their advocacy rotations, a collaborative effort led by Lisa Kessler and Debbie Requesens, PhD, the director of Jump Start program in Penn's Orphan Disease Center, brought them face-to-face with the families Dr. Chung works so hard to help. We've shared their thoughts below.

Our paths cross with Dr. Chung in other ways too. She serves as the representative for the Warren Alpert Foundation on the Advisory Committee for the Alliance for Genetic Counseling Fellowship, which is funded by a generous grant from the Warren Alpert Foundation.





I've followed Dr. Wendy Chung's work for years—first through her leadership in newborn screening and later through her groundbreaking efforts in rare disease research. What's always stood out to me is her unwavering focus on impact: she doesn't just generate knowledge, she brings it directly to families. Her work embodies what it means to do boots-on-the-ground translational research, and she's set a standard for what patient-centered science can look like.

This semester, I've been working with the US MEF2C Foundation for my advocacy rotation—an organization Dr. Chung actively partners with to support families facing MEF2C-related disorders. Seeing her speak in person at Penn was incredibly meaningful. I could never hope to have the scale of impact she's had on the field, but I left feeling more committed than ever to building a career rooted in that same ethos: science in service of families. Caitlin Findlay, UPenn MSGC student, Class of 2027



For my advocacy rotation, I worked with the NEHI Research Foundation. Neuroendocrine cell hyperplasia of infancy (NEHI) is a rare lung condition in which the underlying genetic predispositions have not been well-described thus far. Dr. Wendy Chung is spearheading a genetic research study for the NEHI community, and I had the opportunity to hear her speak to NEHI families about the study at a recent webinar. I was greatly inspired by her approach in talking with these families and her immense dedication to the research. Navigating genetic research can be challenging and there can be many uncertainties, but Dr. Chung spoke with honesty and answered questions with grace, which was a meaningful takeaway for me. I was glad to have the opportunity to hear Dr. Chung speak here at Penn in a different context. The work she has done and continues to do for so many conditions and communities is so impactful, and it was wonderful to catch a glimpse of that work through her seminar.

Emma Cohen, UPenn MSGC student, Class of 2027



This semester, I had the opportunity to work with the CSNK2A1 Foundation as part of my advocacy rotation. The foundation supports families affected by Okur-Chung Neurodevelopmental Syndrome (OCNDS), a rare condition named in part after Dr. Wendy Chung, who played a key role in its discovery. I was excited to learn that Dr. Chung would be speaking at Penn, and it was fascinating to hear her discuss her approach to discovering genes like CSNK2A1. Although OCNDS is rare and much remains unknown, my rotation experience made it clear how meaningful a diagnosis can be for families. Dr. Chung and other researchers have hosted webinars and have developed informational resources to help guide these families, and the OCNDS community is incredibly close-knit and motivated. I really enjoyed working with them this semester to help create an educational video and a few handouts for their website.

Anna Madden, UPenn MSGC student, Class of 2027

Expertise on Display at the GCEA Meeting

In addition to her active roles on the DEIJ and Education Committees at the Genetic Counselors Educators Association, Associate Program Director Lisa Kessler was also invited to share her expertise in two talks at the organization's annual education conference last summer. She participated in panel discussions with other genetic counselors, one on working with standardized patients and the other on Telesupervision in GC education, both critical topics not only post-COVID, but with the increasing number of genetic counseling programs limiting the availability of traditional clinical placements.

Lisa's research on standardized patients in clinical training began at Arcadia University and continued at UPenn, in collaboration with Denise LaMarra, MS, CHSE, and the Penn Medicine Standardized Patient Program, Ian MacFarlane, PhD, Kathy Valverde, PhD, LCGC, and program alum Melissa Heller ('20), now a genetic counselor with the Cancer Risk Evaluation Program at Pennsylvania Hospital. The work was published in the <u>Journal of Genetic Counseling</u>, where it was a top cited article for 2021-2022.

The telesupervision research was conducted at the University of Pennsylvania with then student Michaela Mundt as part of Michaela's thesis research into the use of telesupervision during the height of the COVID pandemic, and it too resulted in a publication in the <u>Journal of Genetic Counseling</u>. Lisa explained that "we learned a great deal during that time which has shaped the way that many students continue to be supervised for clinical education today. Michaela graduated in 2021 and is a valued supervisor for the MSGC program, all the way from California."

It is an amazing opportunity for our students to work with supervisors outside of our geographic area with telesupervision. and Lisa shared this message for program graduates:



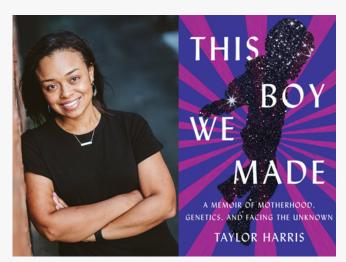


Clinical supervisor is now available for alumni potentially interested in working with students, even if they aren't in the local area. Telesupervision has broken down geographic barriers, and we are always looking for more supervisors to join our community!

Lisa Kessler



This Talk She Gave: Author Back by Popular Demand



Author Taylor Harris

Taylor Harris, the author of *This Boy We Made: A Memoir of Motherhood, Genetics, and Facing the Unknown*, visited the Genetic Counseling Summer Interns on Friday, July 25th, the final day of their six-week internship. Eleven interns, undergraduate students with interest in genetics, from five different sites read the book earlier in the summer and joined via Zoom to hear more about Taylor's experience as the mother of a son with troubling symptoms but no clear diagnosis who fights for answers and the support her son needs from health care providers and the education system, all while struggling with her own

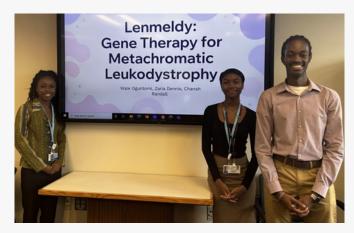
doubts about the choices she makes and with the inequities built into the health care system.

Taylor was back by popular demand following the book discussion last summer that one intern called the highlight of her internship and that led Taylor to comment "What an amazing crew! Those students really encouraged me." The UPenn MSGC program first connected with Taylor in spring 2022, just months after the publication of her book, when she joined us via Zoom for a powerful book club discussion. She calls herself "an open book," and the interns commented on exactly that again this year. They appreciated how honestly she shares her feelings – pain, frustration, doubt, fear, hope and joy – both in writing and in person. She accepted every question appreciatively and answered every question thoughtfully and thoroughly, a literal model of what we can learn when we listen, a critical skill for genetic counselors.

She saves some of her highest praise for Shelly, the Genetic Counselor at the University of Virginia who she met early on in her son's diagnostic odyssey, and with whom her family is still in contact. "She never made me feel small, or that my questions were too 'out there," Taylor said. "She always made space for nuance and the unknown." That allowed her a place for hope, and, over time, to understand that the most important thing is that her son feels loved and "realized" (his words) every day.

The Alliance Summer Scholars Program is led by Lisa Kessler and Shama Khan, MPH, MS, LCGC, Assistant Director/Assistant Teaching Professor at Rutgers University Genetic Counseling Master's Program, the AGCF project site coordinator for the Rutgers MSGC program, and a program alum from our Class of 2014. The summer program is part of the Alliance for Genetic Counseling Fellowship, funded by a grant from the Warren Alpert Foundation. Administered by the UPenn MSGC program, the grant also supports the summer programs at four other MSGC programs: Boston University, Rutgers University, Sarah Lawrence College, and the University of Maryland.

Interns attend Genetics Rounds, case conferences, and lectures, shadow in lab and clinical settings, work on special projects, network, and connect with current students. Dozens of people at each site are involved in creating opportunities and educational experiences for the interns, and we are so appreciative of their efforts. Since beginning in 2022, more than 35 undergraduate students have participated, and many have now applied to and enrolled in master's programs.



2025 UPenn Genetic Counseling Interns presented their research project the last week of the summer internship. From left, Chanah Randall, (Rosemont College, '26), Zaria Dennis (Chestnut Hill College, '26), and Wale Oguntomi (Haverford College, '27).

A Network of Caregivers - A Network of Alumni



66 It was the best case!"

That's what Kathy reported one Tuesday morning in December 2023 as she returned to the office from Genetics Rounds at CHOP. Avi Anantharajah, MS, LCGC, and Jackie Cappadocia, MS, LCGC, genetic counselors at the Basser Center for BRCA, had presented a complex cancer case and described in detail the process of diagnosis and medical management. Equally significant, they outlined the critical role of collaborative work across institutions, between clinical and laboratory based genetic counselors, and with patients themselves.

The 45-year-old breast cancer patient was first seen by genetic counselors Corinne Fillman and Briana Marmelstein at the Cancer Risk and Genetics Program at St. Luke's University Health Network in Bethlehem, PA. Based on several factors, they suspected Diffuse gastric and lobular breast cancer syndrome, but genetic testing revealed a CDH1 c.833-9 C > G (intronic) VUS, which did not meet the current International Gastric Cancer Linkage Consortium clinical practice guidelines for gastric surveillance recommendations. Nevertheless, given her history and the uncertainty of the variant, multiple cancer specialists recommended an endoscopy, and the resultant biopsy revealed "a focus of signet ring cell carcinoma."

Samples were sent to Ambry Genetics for RNA sequencing and further analysis by genetic counselor Carrie Horton. Their findings, combined with the patient's history and data collected from a similar case, provided enough evidence for "reclassification of the variant from uncertain significance to likely pathogenic." This change will guide care for future patients with suspected diffuse gastric and lobular breast cancer.

An article on this case, "Combining clinical and molecular characterization of CDH1: a multidisciplinary approach to reclassification of a splicing variant," was published in Familial Cancer and written by Fillman and Marmelstein at St. Luke's, Anantharajah and Bryson Katona, MD, PhD, at Penn, and Horton from Ambry Genetics, with other colleagues. They emphasize both the profound impact multidisciplinary collaboration can have on variant resolution and the importance of having complex test results reviewed by genetics professionals to ensure appropriate interpretation in the clinical setting.

Resolution of a VUS can provide straightforward, variantbased guidance for clinicians and patients, and this case highlights the importance of collaboration between clinicians and reporting laboratories in VUS resolution. There are currently no standardized methods to facilitate longitudinal communication of clinical data between clinicians and laboratories, however such collaborative efforts can yield immense benefits to clinical care and result in potentially lifesaving interventions.

A critical piece that facilitated this collaborative network and made Kathy so excited: they are all graduates of the UPenn/ Arcadia MSGC program: Corinne Fillman ('08) and Briana Marmelstein ('20) at St. Luke's, connected with Avi Anantharajah ('21) at Penn and Carrie Horton ('08) at Ambry Genetics.

"It is always rewarding to see our graduates accomplishing great work in clinical settings, research, advocacy, and leadership," Kathy said. "This case is a reminder that they are not just skilled individuals, but part of a large network of 396 program alumni in innumerable settings who can consult, support, and inspire each other throughout their careers."





Fillman C, Anantharajah A, Marmelstein B, Dillon M, Horton C, Peterson C, Lopez J, Tondon R, Brannan T, Katona BW. Combining clinical and molecular characterization of CDH1: a multidisciplinary approach to reclassification of a splicing variant. Fam Cancer. 2023 Oct;22(4):521-526. doi: 10.1007/s10689-023-00346-z. Epub 2023 Aug 4. PMID: 37540482.

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We want to help CURE FA!

Every October since 2012, our very first year volunteering at rideATAXIA Philly, MSGC students have been up bright and early to direct traffic, hand out water, cheer on riders, staff the registration table and more, at this annual event to benefit the Friedreich's Ataxia Research Alliance (FARA), a national, non-profit organization dedicated to curing Friedreich's ataxia (FA), a debilitating, lifeshortening, degenerative neuro-muscular disorder that is usually diagnosed in childhood, through research. (We're proud to claim FARA CEO Jennifer Farmer, a graduate of our program's very first class in 1997.)

We've been there because every September, Kyle Bryant, spokesperson for FARA and founder and director of rideATAXIA, comes to campus to meet the students and share his story and his unflagging determination to cure FA. Though diagnosed with FA at 17, he has captained a team of riders for Race Across America, one of the most challenging contests on earth, and, in 2024, formed another team to reach the peak of the highest road, Umling La Road at 19,300 feet, in the Himalayan Mountains, all to raise awareness about FA. *The Ataxian*, a documentary film about the cross-country ride, was released in 2015, and *The Highest Road*, a new documentary about the incredible climb, is in development. He cohosts a podcast called "Two Disabled Dudes" and wrote a book titled *Shifting into High Gear*. All the while, he is the irresistible force bringing people together to raise money, ride and connect at rideATAXIA events around the country, which together have raised over \$11 million for FA research.

Other members of the FARA team join him on campus to share the struggles faced by those with FA, explain the efforts being made to improve awareness and treatment, and the progress toward finding a cure. After the class, the students eagerly sign up to volunteer at the fundraiser.

The past year, it all came together for what the students called "an amazing session." Kyle was joined by Kellyn Madden, MS, LCGC, the Patient Engagement Manager with FARA (and a program alum from the Class of 2023); McKenzie Wells, MS, LCGC, Clinical Research Coordinator in the Friedreich's Ataxia Program at CHOP (another alum, from the Class of 2011); and Elizabeth Mercado-Ayon, a PhD candidate at the University of Pennsylvania in Cellular and Molecular Biology, where she investigates the circuits of the cerebellum in FA mouse models in the lab of David Lynch, MD, PhD, director of the FA program at CHOP, and a powerhouse of FA research. Each person shared their unique expertise with the students, and, no surprise, we had a great turnout at the event later that fall. We're looking forward to their visit this fall and to volunteering at rideATAXIA Philly 25. You can learn more about FA, FARA and rideATAXIA at https://www.curefa.org/.



In the classroom, above, and some of our volunteers at rideATAXIA Philly 2024, below.





22q 360°

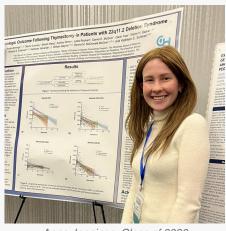
The MSGC program has an all-year-round relationship with CHOP's 22q and You Center. Students attend clinical placements at the Center, and they also volunteer at the annual 22q and Boo and 22q at the Zoo. Director Donna McDonald-McGinn, MS, LCGC, lectures in first- and second-year courses on a wide range of topics, from 22q11.2 deletion syndrome and craniofacial disorders to conducting chart review research and preparing conference abstracts.

Donna has been a champion of the MSGC program since the days of its inception in the mid-1990s at Arcadia University. One of the first genetic counselors hired at CHOP, she has built an international reputation as an expert in 22q11.2 deletion syndrome, and she has brought countless students along with her. Perhaps her most notable contributions to the MSGC program are the 20 thesis research projects she has mentored and the many presentations and publications that have resulted.

Our students graduate with exceptional knowledge of this multi-system condition, and they carry that knowledge with them into their careers.



MSGC students at the Philly 22q at the Zoo in May 2023



Anne Jennings, Class of 2026, presenting at the PAGC Conference



Sarah Donoghue,'19, Jen Borowka '24, Kathy Valverde, Daniel McGinn '23 & Bekah Wang '22, presenters at the 13th Biennial International 22q11.2 Scientific Conference in Portugal, 2024.

A Voice for Genetic Counseling



Evan Hathaway, MS, LCGC

One of the highlights of the first semester in the MSGC program at UPenn is the Genetic Counseling Panel, held as part of the Introduction to Genetic Counseling course. For the culminating event of the course, we invite a panel of genetic counselors to share their professional journeys, career goals, and aspirations with the first-year students.

In December 2024, panelist **Evan Hathaway**, MS, LCGC, spoke about his role as a genetic counselor at the Beckwith-Wiedemann Syndrome and Overgrowth Genetics Clinic at CHOP. Evan, an alum from the Class of 2017, provided valuable insights into his experience working in this highly specialized field.

Through his work, Evan has had numerous unique opportunities, including participation in research studies and involvement in the BWS Conference at CHOP. He was also invited to coauthor new guidelines on the diagnostic approach to lateralized overgrowth on behalf of the ACMG Professional Practice and Guidelines Committee. The guidelines, titled "Isolated lateralized overgrowth and the need for tumor screening: A clinical practice resource of the American College of Medical Genetics and Genomics (ACMG)," were recently published in Genetics in Medicine. The senior author of this publication is Dr. Jennifer Kalish, the Director of the BWS and Overgrowth Program at CHOP and Evan's mentor. We are so proud of Evan for representing genetic counselors on this panel and participating in this impactful work, and we were excited to have Evan join the panel and share his expertise with the next generation of genetic counselors.



Spotlight Q & A:

A Model for Mentoring GC Student Research

continued from page 7

How do project ideas get started — do you keep a running list of research ideas or do you work with interested students to develop projects?

Bryson Katona, MD, PhD (BK): There are so many incredibly interesting research ideas in the gastrointestinal (GI) cancer genetics field! Most of these research ideas develop directly from questions that result from seeing patients who either carry a hereditary GI cancer risk syndrome or who are at risk for carrying such a syndrome. I do keep a running list as these questions arise throughout the year, and if a student is interested in working on one of the questions, I then work directly with the student to develop and refine the research question and the approach that we will have to take to answer it.

How do you identify a project or piece of a larger project that can be accomplished by a student in the limited time they have? How do you assess what a student should be able to accomplish by graduation?

BK: Carefully picking a feasible project is by far the most important thing that can be done to facilitate a genetic counseling student having a successful thesis experience. Given the limited timeframe in which GC students need to complete their thesis, selecting a project that already has IRB approval or a project where IRB approval can be obtained efficiently is imperative, as that way the student can start doing meaningful work on the project as soon as possible. Having worked with many students over the years, I have a good sense of what a student can accomplish for their thesis. Students themselves may underestimate how much time classes and clinical rotations take, so it is important to discuss the level of effort that will be required with the student early on in the project.

Do you/how do you divide up roles on the mentorship team? Who monitors student progress, provides access to patient records, connects students with educational background, statistical support, editorial feedback, etc.?

Jacqueline Cappadocia, MS, CGC (JC): When dividing roles on a mentorship team, it's important to take a personalized approach that recognizes each student's current strengths while also identifying opportunities for growth. The goal is to create a supportive environment where students can gain confidence by tackling new challenges, balanced with appropriate guidance and resources for areas beyond their current expertise.

Typically, the primary thesis advisor provides broader direction and expertise for how the project is designed and implemented. The thesis advisors work together to monitor the student's overall progress, ensuring that milestones are met and providing regular feedback. Access to patient records and sensitive data is often managed by a designated team member familiar with institutional protocols, ensuring compliance with privacy regulations etc.



Samantha Williams, '22, and Bryson Katona at the CGA-IGCC Conference in 2022



Spotlight Q & A: A Model for Mentoring GC Student Research

continued



Have/how have you refined the mentoring process over time as you've learned from your experiences?

Derek Mann, MS, CGC (DM): I have been able to get comfortable in more of a guidance and support role compared to a lead role that I would have with my own projects. It has been great to see how the students can thrive knowing that we trust and believe in them to be the lead on their project, while also knowing that we are always there to answer questions, give feedback and provide help when needed. I have seen that this leads to a great working relationship between the mentors and students, and provides the students with confidence and pride in their work.

JC: I've learned important lessons about finding the right balance in mentorship. It's crucial to be hands-on to provide strong support and guidance, helping the student feel well-supported, while not overstepping or taking over their work, allowing them space to grow and develop independence.

I've found that this balance requires ongoing adjustment—knowing when to step in more actively, such as during challenging moments, and when to step back to encourage autonomy. This awareness helps foster a productive and positive mentoring relationship that supports the student's learning and growth.



What recommendations would you give to researchers who are just starting to work with student researchers?

Jessica Long, MS, CGC (JL): Between the graduate coursework, clinical rotations and thesis responsibilities, the semesters fly by, so regular communication is key! For each student thesis project, thesis mentors Dr. Bryson Katona and the participating genetic counselor(s) meet with the students every other week during a standing 30-minute meeting. We're also in contact via e-mail between meetings as needs arise. By committing to bi-weekly meetings, the team can keep the project moving forward in a timely manner and ensures everyone is aligned regarding project needs and next steps.

JC: It is also crucial to help them manage their workload effectively, especially since they have many other clinical responsibilities on during their second year. One key recommendation is to set smaller, attainable goals with intermediate deadlines throughout the research process. It can be as simple as "try to review X amount of charts by our next meeting." This approach encourages consistent effort over time, preventing the accumulation of last-minute work that can become overwhelming as the final program deadline approaches. Breaking the project into manageable milestones not only helps students stay on track but also builds their confidence and promotes steady progress, resulting in higher quality outcomes and less stress for both students and mentors.

DM: Finally, I would recommend always providing encouragement and highlighting the good work the student has completed. For some of these students, this will be the first time they are working on a research project, while the others will likely have only some experience. The world of IRBs, creating the project and data collection can be overwhelming, and students can feel lost not knowing how their work is measuring up. Providing a "good job" or commenting on something you like that added to the project can go a long way and give the student the boost they need knowing that they are on the right track.



Sam Levy, '24, and Bryson Katona, MD, PhD, at the Perelman School of Medicine Master's Student Colloquium in 2024.



Spotlight Q & A: A Model for Mentoring GC Student Research continued



After a student graduates, how do you communicate with them to encourage them to keep working toward publication?

JL: By graduation, our students have submitted their thesis for MSGC program review and their degree requirements. However, we've found the Penn MSGC students are highly motivated to present their work at relevant conferences as posters or invited presentations and to prepare manuscripts for publication! This is in part because they have typically selected these projects as relevant for their own professional interests, but also because our students are typically very eager to see their work directly inform and advance clinical topics related to hereditary cancer genetic counseling and testing!

Because manuscript preparation and submission for publication is typically the end goal, efforts have consistently been made during the student's graduate training to develop their thesis with this in mind. Post-graduation, we continue meeting as needed until submission is complete, and we're thrilled that our trainees have had a very high manuscript acceptance rate! Also, since we work hard to establish relationships with our students that are positive and supportive, the completion of a project is bittersweet when our regular meetings end – but we still look forward to seeing many of our past students at future professional meetings and as colleagues!

Jessica Ebrahimzadeh, **MS**, **CGC (JE)**: It is helpful to identify the academic journal they plan to submit the manuscript to prior to completing their final thesis assignment with the support from their mentors. Each journal has specific format requirements, which can also be used for their final thesis assignment for the MSGC program to help reduce extra work.

?

Have/how have students helped your research progress?

JL: It has been tremendously helpful to have Penn's MSGC students dedicate their time and skill sets to helping critically develop projects, collect data and draft abstracts, posters and publications. The students are well-trained by the MSGC program to support all aspects of the research projects, and many come into graduate school with prior research experience. These have been excellent opportunities to further investigate clinical questions or scenarios that we've encountered during our daily work in the hereditary cancer genetics clinics. And, involvement in student research has enabled me to learn and professionally grow right alongside our mentees, thanks both to their efforts and to the expertise and mentorship Dr. Katona provides for these projects.

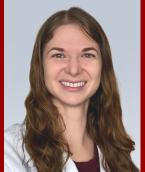
JE: Echoing Jess; I've learned invaluable research and project management skills from working with Dr. Bryson Katona. As a genetic counselor with minimal research training, I appreciate the ability to collaborate and learn from Dr. Katona alongside the MSGC students. It gave me the ability to more independently mentor a student thesis project for the most recent graduating class.

Thank you!

Bryson Katona, Jess Long, Jes Ebrahimzadeh, Jackie Cappadocia, and Derek Mann for sharing your knowledge and experience with us!











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Class of 2022



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