INVITAE

THE HEART OF GENETIC COUNSELING AWARD 2023

The Heart of Genetic Counseling AWARD

Presented by INVITAE National Society of Genetic Counselors

Genetic counselors play a unique role as patients and families navigate health challenges that may have significant, lasting consequences. They bring extensive knowledge of genomic medicine and heartfelt empathy to every step of the patient's journey. As the growth of genomic medicine continues to impact our medical care, genetic counselor expertise is invaluable. Regardless of their work setting, genetic counselors advocate for the use of genetic information in all areas of health care and Invitae is honored to celebrate them.

— Ken Knight, President and Chief Executive Officer, Invitae

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Presented by

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National Society of Genetic Counselors

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Dedication

THE HEART OF GENETIC COUNSELING AWARD celebrates the heart that genetic counselors bring to the front lines of genetic medicine, as they help patients through life-changing decisions for themselves and their families.

Presented by the National Society of Genetic Counselors and Invitae, the Heart of Genetic Counseling Award honors distinguished genetic counselors who have made a difference in the lives of their patients by using the combination of clinical excellence and human compassion that defines the profession. The stories in this book are a testament to genetic counselors' commitment to their patients and to the life-changing impact they have on the people they touch.

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The Hearing Heart

Patrick L. Wilson
MMSc, MS, LGC, CGC

Clinical Assistant Professor
University of Oklahoma Health Sciences Center/Department of Obstetrics and Gynecology
Section of Maternal-Fetal Medicine

Photograph by Mallory Yockel

WITHIN THE HEART OF EVERY GENETIC COUNSELOR is an ear of empathy. This ear hears the patient and interprets what is said so that the heart can express the empathy the patient needs during an emotionally trying time.

Our patients do not come to us because something wonderful happened to them. They come seeking answers to health concerns that adversely affect them or a family member. They expect to talk to an expert about a health problem, testing, or medical procedures. Oftentimes, they do not expect the expert to listen to them, to be empathetic.

The empathetic heart of genetic counselors distinguishes us from other healthcare providers. We allow our hearing hearts to dictate our responses to each patient. We see patients and family members as individuals, not as conditions. We hear the whole person. We hear the outside factors that prevent them from attending appointments, obtaining tests, or receiving treatment, and address them with empathy, not shame.

As you read each vignette in this year's Heart of Genetic Counseling Award book, you will see how counselors' hearts brighten their patients' perspectives. You will see how counselors' hearts allow their patients to be people and not diagnoses. Lastly, you will see how the ears within counselors' listening hearts show their patients that genetic counselors truly have hearing hearts.

We allow our hearing hearts to dictate our responses to each patient. We see patients and family members as individuals, not as conditions. We hear the whole person.

To illustrate the hearing heart, I recall a recent experience I had with a parent who had two daughters with Gaucher disease. Both girls were scheduled to receive enzyme replacement therapy but at different times. However, because the timing of the infusions was inconvenient to the mother, one of the girls would miss her treatment. Many people on the care team were upset due to the cost of the medication and the perceived lack of concern by the mother.

Because I speak and understand little Spanish, I asked a Spanish-speaking physician to accompany me on a visit to the patient's house on the day the girls were receiving their infusions. The mother explained to us that the infusions were on alternating schedules: The younger, more severely affected girl received treatments each week in the afternoon, and the older, less severely affected girl received biweekly treatments in the morning. The mother worked nights and slept in the mornings, so it was difficult for her to stay awake on Thursdays when both girls received treatments. Often, the younger daughter would miss her infusions. The home health agency did not want to adjust the schedule and perform the infusions solely on Thursday afternoons.

After reporting the reason behind the missed infusions to the physicians, we were able to move the infusions to our genetics clinic. We were able to infuse the girls on a date and time that was convenient for the family, and they were present for each infusion thereafter.

The above scenario is not uncommon. Outside distractions often prevent our patients from doing what is asked or desired of them. What distinguished me from the rest of the care team was that I took an active interest in the patient. My heart was not hardened by a perceived maternal lack of concern for the seriousness of Gaucher disease and the need for regular infusions to limit its effects. My heart listened to what my ears were hearing, and a solution was found to the problem. The results were that the physicians I worked with were less stressed, the mother was happy someone heard her, and the girls received the treatment they needed to live better lives.

The Winner

VICTORIA

Victoria Suslovitch
Unsung Hero

Victoria Suslovitch
MS, CGC

Ambry Genetics
Aliso Viejo, Callifornia

Nominated by
Kara Boulter

Photographs by
Benjamin Norman

MY 5-YEAR-OLD SON, NOLAN, has a rare disease called KCNQ2 developmental epileptic encephalopathy. Nolan has many struggles due to this diagnosis; in addition to seizures, he is nonverbal, nonambulatory, and dependent on a gastrostomy tube. Nolan has been diagnosed with numerous other ailments, including spastic quadriplegic cerebral palsy, cortical visual impairment, gastrointestinal dysmotility, esophageal reflux, chronic lung disease, and severe global developmental delay.

Nolan's rare disease brought us to the Children's Hospital of Philadelphia (CHOP), where we are privileged to have an extraordinary team of doctors and a wonderful genetic counselor who are intimately familiar with his disease. Through CHOP, we were led to the neurogenetics team at Boston Children's Hospital (BCH). At BCH, we met another amazing genetic counselor, Victoria "Tori" Suslovitch. Tori works with Dr. Tim Yu's lab, which works with other research groups to develop genetic treatments for extremely rare conditions.

Science takes time, and KCNQ2 has proven to be more challenging for the team than anticipated. However, I believe the only way to give Nolan the opportunity to live his best life and significantly reduce his suffering is through genetic treatment.

The waiting and the unknown have been very difficult for my husband, Tom, Nolan's 6-year-old brother, Thomas, and me. I am obsessed with saving Nolan. Tom is often overwhelmed with all that KCNQ2 has stolen from both his sons, and Thomas is angry and sad that his brother is incapable of being the built-in best friend he yearns for. One would think as the years go on, our family would have settled into a routine, that with time there would come understanding, acceptance, and normalization. While we are able to enjoy small moments and some days, the raw truth is with time our pain deepens, and Nolan's challenges are more apparent.

Tori's unique dynamic as a bereaved sister and genetic counselor gives her the ability to give herself to families in a way most counselors cannot. She connects with siblings as a peer and professional and understands parents through the eyes of her mother.

Against this backdrop, Tori emerged into our lives. She graciously answers my countless emails badgering her for updates and shows remarkable patience when confronted with any number of agonizing questions and emotions I put on her. Tori is readily available, and with each response from her, a bit of calm washes over me. But Tori's impact goes beyond the support she offers me and extends to what she does for Thomas.

As I came to find out, Tori knows all too well what Thomas is going through and how he feels. In one of our many conversations, Tori shared that she had a sister with special needs who passed away.

She has helped me understand and navigate Thomas' anger amid his growing awareness of all the things his brother can't do, and the pain Nolan must endure. Thomas has witnessed critical incidents that no 6 year old should experience, including his brother's seizures, mucus plugs being ripped from his brother's lungs to clear his airway, and 911 calls. He has had to take a backseat to his brother more times than is fair.

Tori shared her own thoughts and emotions on her similar journey as a rare disease sibling. She spoke about what coping mechanisms she developed and shared some that her mother used to help her as a child. Tori has provided resources and tools for Thomas. She has taught me how to communicate with him and to give him opportunities to express himself by any means comforting to him.

Tori has shown me that Thomas' feelings — his anger, sadness, jealousy of healthy siblings, and sensitivity — are all normal. She reassured me that it is okay to be open and honest with him and to validate his emotions. She has also taught Thomas how to utilize healthy coping mechanisms.

Tori helped me believe that Thomas will be okay, that he can grow up to be a happy, healthy, loving, and empathetic adult, just like Tori did.

Tori's unique dynamic as a bereaved sister and genetic counselor gives her the ability to give herself to families in a way most counselors cannot. She connects with siblings as a peer and professional and understands parents through the eyes of her mother.

Tori repeatedly replies to my many late-night emails and never ignores me. Tori's responses always make me feel heard. Often, I feel lost and out of control, unable to protect my baby. I can't heal him. I can't stop his pain. It can be the loneliest and guiltiest of feelings, but sometimes just knowing I'm being heard in the moment is what I need.

Tori's impact goes beyond her comforting and responsiveness. She has taught me how to be the mother Thomas needs and helped me see the world through his eyes. She gave me something money can't buy — faith that Thomas can and will be okay and validation that I am the mother both my sons deserve.

Tori is an unsung hero for all rare-disease siblings and families. She is a guiding light through an unknown, frightening medical journey — a road she is all too familiar with.

CLOCKWISE FROM UPPER LEFT: KARA, TOM, VICTORIA, NOLAN, THOMAS

“Tori is an unsung hero for all rare-disease siblings and families. She is a guiding light through an unknown, frightening medical journey — a road she is all too familiar with.”

The Finalists

Leslie Walsh Cyprych "Just Be Mom"

Leslie Walsh Cyprych
MS, MPH, CGC

Children's Hospital of Pittsburgh
Pittsburgh, Pennsylvania

Nominated by
Tracy Fisher

Photographs by
Rob Larson

IF YOU WERE TO ASK ME the definition of advocacy, it wouldn't be a description. It would be one word: Leslie. I have never met such a professional and knowledgeable person in our daughter's journey. When it comes to navigating the healthcare system, Leslie can literally do magic — expediting our appointments, helping us with billing issues, or saying exactly the right thing at the right time.

My husband and I met Leslie on the worst day of our lives when our 11-month-old daughter, Brie, went from being a normal, healthy baby to a child that had something very wrong with her. For the first 11 months of her life, Brie hit all of her milestones, sometimes even earlier than expected. She was able to stand up and then eventually take a few steps. But around 11 months old, Brie stopped smiling. She began sleeping more and was hard to wake up. After about a week of this, I scheduled a telemedicine appointment with her pediatrician. I was worried enough to make an appointment, but not worried enough to take her into the doctor's office.

As soon as her pediatrician saw Brie's flaccid demeanor, he had me bring her in immediately. Multiple MRIs of Brie's brain revealed the probability of her having a degenerative disease that would leave her severely impaired, both physically and cognitively. On that devastating day, Leslie and the genetics director introduced themselves and discussed our next steps. While other medical staff were present at that meeting — nurses and neurologists — I felt Leslie's confidence. She owned the room. We were heartbroken, but Leslie's presence was calming and reassuring.

As rare as Brie's genetic disease is, I feel like Leslie is just as rare in her field.

Our medical team ordered rapid, whole-exome sequencing, so we could receive a diagnosis in two weeks, not three to six months. Every day without those results, we were dying inside to know what was happening with our daughter. On March 1, 2021, two days ahead of schedule, Leslie met with us to deliver the news. She had prepared a packet that broke down everything about Brie's diagnosis of Aicardi-Goutieres syndrome (AGS), a rare, progressive, and incurable genetic disease. Leslie explained that two AGS specialists were at the Children's Hospital of Philadelphia (CHOP). Rather than have us contact CHOP ourselves, she hopped on her computer during the meeting and did her magic, writing a huge letter of medical necessity to get us into CHOP sooner. Even though, at that time, Brie was relatively asymptomatic, they could tell, based on her brain scans, that the storm was coming. Leslie told us waiting nine weeks to see a specialist was not going to cut it.

Because of Leslie, we received a call from CHOP just 45 minutes later while we were on our way home. She and our medical dream team helped facilitate getting us in to see a specialist within a week. This is important, because two months after Brie's diagnosis, the big regression hit. She couldn't sit up or even hold her head up. Basically, the disease just popped its head out and said, I'm coming for you.

Two years later, Leslie continues to be a huge advocate for Brie, now 3. She has helped expedite appointments at CHOP and our local hospital for Brie's echocardiograms and numerous other tests. She has helped with billing, even calling a genetics lab to sort out costs. When Brie has been hospitalized with neutropenia and other health issues, Leslie will just appear out of nowhere with a cake pop for Brie and a coffee for me. She just makes things happen.

The biggest example of Leslie's compassion that will forever live with me is her words at that first meeting when we learned Brie's diagnosis. She said, "I know you're overwhelmed with everything right now, but we will handle everything on the medical side. You just be Brie's mom. Just be Mom." Those three words have encapsulated this whole journey for us, because it's so scary. I'm not a doctor or a genetic specialist, but when Brie has hard days, or we receive bad news, I always think of Leslie and those three words — "Just be Mom." That is just one of the many "Leslie-isms" that have comforted me along this journey. As rare as Brie's genetic disease is, I feel like Leslie is just as rare in her field.

L TO R: TRACY, BRIE, LESLIE

“The biggest example of Leslie’s compassion that will forever live with me is her words at that first meeting when we learned Brie’s diagnosis. She said, ‘I know you’re overwhelmed with everything right now, but we will handle everything on the medical side. You just be Brie’s mom. Just be Mom.’”

ANDREA

Andrea Gainey
Our Superhero

Andrea Gainey
MS, LCGC

UConn Health
Huntington's Disease Program
Farmington, Connecticut

Nominated by
Susan McGann

Photographs by
Joe Buglewicz

Andrea Gainey by Susan McGann

I HAVE KNOWN MY HUSBAND, JAMES, and his family since 1970, when we met in college. At the time, we did not know Huntington's disease (HD) was in his family. We got married, had kids, and then found out his mother had HD. James' family did not talk about the disease or its genetic factors. At the time, there wasn't a test available because the gene had not been identified. So, we carried on and lived our lives.

Then, James' sister developed symptoms of HD. She died from the disease in 2006 at age 63. Around that time, I started volunteering at the Connecticut Chapter of the Huntington's Disease Society of America, helping to raise awareness and plan fundraisers. That is where I first met Andrea and learned about the UConn Health Huntington's Disease Program, the only state-funded HD program in the country.

The UConn program started in 1997 after a patient had been misdiagnosed with HD and committed suicide. Andrea was the program's first genetic counselor. She has helped generations of families and understands the stigma that surrounds HD, which is why she established free anonymous testing and confidential lab reports early on in the program.

If a person has a parent with HD, he or she has a 50 percent chance of inheriting the gene. Since HD is a degenerative brain disease without a cure, many people don't want to know they have the gene. Only 10 percent of those at-risk of inheriting the disease get tested. And 25 percent of HD-positive people commit suicide.

Last year, Andrea made capes for the HD patients to wear at the walk. The capes said "superhero," because Andrea believes those fighting HD are superheroes. However, Andrea is our superhero too — helping us fight HD every step of the way.

I shared the testing information with James, who had been researching HD, unbeknownst to me. At the time, he was in his mid-50s and did not have symptoms of HD. We decided to make an appointment with Andrea. We were told that we should have long-term care insurance in place before we started the process.

It takes a special person to deal with this devastating disease on a daily basis and stay positive, but that's Andrea. She understands how difficult the decision to test is and knows how to put you at ease. She spent a lot of time asking my husband questions about why he wanted testing and how he would react if the results were negative or positive. After she was convinced, she referred him to a neurologist and social worker to assess his readiness for testing.

After meeting with the social worker and neurologist, my husband and I felt he would be negative since he had no symptoms and was past the age when most patients become symptomatic. We moved forward with the testing.

Three weeks later, the day after Christmas, we met with Andrea for the test results. Unfortunately, my husband had inherited the HD gene. We were shocked. We knew it was a possibility, but even the neurologist was surprised. Andrea was wonderfully helpful and comforting. She gave us support group information and the phone number of the group's leader. She provided clinical trial information and told us there was hope for future treatments. Most of all, she told us she would be by our sides during the journey ahead.

It has been 10 years since we first met Andrea, and she continues to support us and our family. We attend Andrea's monthly support group meetings. Our children have decided not to get tested yet, but they know Andrea is there for guidance and emotional support when they are ready. I know Andrea provides that same degree of care and comfort for other families because I still volunteer in the HD community and have heard from many others about how Andrea has gone above and beyond providing support during difficult times.

Andrea is also considered a leader in the HD community and teaches genetic counselors and other healthcare providers about the disease. She has lectured at conferences and events to promote awareness of HD. She is a resource for HD families, not just in Connecticut but also around the country.

She also is a great supporter of our local chapter of the Huntington's Disease Society of America. For years, UConn did not sponsor our fundraising walks. We asked but there was always an excuse. A few years ago, there was a change in leadership and Andrea jumped on the opportunity to press for sponsorship for our walks. Since then, UConn has sponsored our walks every year.

Last year, Andrea made capes for the HD patients to wear at the walk. The capes said "superhero," because Andrea believes those fighting HD are superheroes. However, Andrea is our superhero too — helping us fight HD every step of the way.

L TO R: SUSAN, ANDREA

“It takes a special person to deal with this devastating disease on a daily basis and stay positive, but that’s Andrea. She understands how difficult the decision to test is and knows how to put you at ease.”

The Nominees

Erin Campbell
Living Without Fear

Erin Campbell
MS, CGC

University of Tennessee
Medical Center/University
Genetics Oncology Clinic
Knoxville, Tennessee

Nominated by
Jennifer Rolen

Photographs by
John Black

MY MOTHER'S SIDE OF THE FAMILY has a history of women who have had ovarian or breast cancers. Two of my aunts died of cancer, one at age 42 of colon cancer and one in her 50s of breast cancer. By the time I was a teenager, my mother had fought and beaten vulvar cancer and had a tumor removed from the tip of her nose. All of these experiences led my mother to genetic testing. Following a positive test for the BRCA2 gene mutation, my mother had a preventive hysterectomy and mastectomy. I saw, in great detail, how difficult and painful these decisions and surgeries were for her.

After the birth of my second daughter, I had my gallbladder removed due to gallstones from pregnancy. After my surgery, my mom called and asked what the pain from the gallstones was like. She was scared because she was having similar pain.

My mother wasn't able to get an appointment with her doctor quickly, so she went to the emergency room. It was there she was told she had stage 4 biliary cancer. We were shocked because she had a physical three months before that revealed no abnormalities. My mom encouraged me to get genetic testing, as did my gynecologist and primary care doctor. However, I was terrified to do it. I didn't know if I wanted to find out whether I had won or lost the "genetic lottery."

My first appointment with Erin was in March 2021 and was over the telephone because of COVID-19. Because I was so scared, our first conversation was heavily punctuated with my crying jags as I tried to process my conflicting emotions. I poured my heart out to Erin and told her that I always believed that I would die young of cancer. My mom's recent diagnosis of an incredibly aggressive and always fatal cancer was just another example. I felt so unstable and apologized to Erin for crying. I'll never forget what she said: "It's okay. This is why they call me a genetic counselor and not a genetic tester."

Even before I got my results, I was aware of the profound effect Erin had on my life. I don't think I would have had the courage to get tested without Erin's unwavering support. Without her care and compassion, I would still be living in fear of the unknown.

After I calmed down, Erin explained that since the BRCA2 mutation does not usually cause the type of digestive cancers that were seen in my family, she suspected that another mutation was at work. She discussed what some of those mutations were and what preventive measures would be recommended. This information doubled my fears as some of the mutations (such as the one for Lynch syndrome) were scarier to me than BRCA2. I was beside myself. Erin recognized the stress I was under and, more importantly, empathized with me. She was so easy to talk to and never pressured me. We decided I needed time to process the information and to revisit in October.

We talked again on October 4. I remember this date because it was two days after my mom died. I was an emotional wreck, but Erin has a wonderful way of making me feel safe, even when we talk about terrifying topics. We spoke about what just happened and what it meant for my future. We covered an array of topics from testing and recommendations to my near-phobic feelings about being tested and my fears for my children and my family. We discussed how my mom took all the preventive steps that were recommended by her health team, which were extensive, painful, and expensive, and yet still died of a cancer that came out of left field. We agreed that I needed further counseling before I was ready for testing. It took two more appointments and the better part of a year before I was able to do the tests Erin recommended.

Up to this point, all of our appointments had been on the telephone, but I was given the option for an in-person meeting to hear my results. I wanted to meet the person who had walked me down such a difficult road and who I knew, beyond a shadow of a doubt, would be with me wherever the results of my tests would take me.

The day of my appointment, I was so anxious I could not breathe. Erin walked in and we talked a little bit. I say "little" because Erin was too excited to hold back the news that all my tests came back negative! I broke down in uncontrollable sobs. It was the news that I spent so many years hoping for, but not believing it could happen. I felt free of my family's curse, and more importantly, that my daughters would be too. Before we parted, I asked Erin if I could give her a hug, and she said, "I was hoping you would ask."

Even before I got my results, I was aware of the profound effect Erin had on my life. I don't think I would have had the courage to get tested without Erin's unwavering support. Without her care and compassion, I would still be living in fear of the unknown.

L TO R: ERIN, JENNIFER

Jamie Dokson
Beacon of Light

Jamie Dokson
ScM, LCGC

Kaiser Permanente Southeast
Medical Group
Atlanta, Georgia

Nominated by
Meleah Brown

Photographs by
Lynsey Weatherspoon

Jamie Dokson by Meleah Brown

MY JOURNEY TO MOTHERHOOD was a difficult process. My first pregnancy ended in a miscarriage followed by an endometriosis diagnosis and surgery to save my left ovary before our son Everett was born. He was 3 years old when we started investigating in vitro fertilization (IVF) as our next step to having another baby. We chose genetic carrier screening, and the results changed the course of my life. I was a carrier of Duchenne muscular dystrophy/Becker muscular dystrophy (DMD/BMD). We had no family history, but Everett had displayed early signs of the disease and had been in physical and occupational therapy since he was 4 months old.

We met Jamie after we received the results and were looking for guidance on next steps. My new carrier status felt like a tsunami that came with multiple fronts. I found it difficult to endure the grueling toll of managing all the new parts of my life and felt lost.

Although DMD/BMD was not her expertise, Jamie bridged the gaps, found answers, and helped us manage these multiple fronts. The "me" front, which involved following up on my increased risk of developing cardiomyopathy on top of my existing bicuspid aortic valve. Jamie encouraged me to work with my cardiology team to make sure I was taking care of myself first.

The "IVF" front involved potentially testing my embryos for the mutation, on top of the regular IVF process. Jamie set us up with a genomics company to develop a test specifically for our situation and worked directly with the fertility clinic to finalize the IVF options and screen our embryos.

The "Everett" front was getting my son tested and finding a specialist, which I learned could have a wait time of six months to a year. Jamie worked with our pediatrician to have Everett tested while we waited to see a specialist at Children's Healthcare of Atlanta.

And finally, "my extended family" front that involved telling my family about this new genetic information. Jamie contacted my family members to offer follow-up testing and assisted them with that process.

When Everett's test results came back positive for DMD/BMD, we thought we'd have to wait six months to see a specialist to get an interpretation. But Jamie took our information to one of her colleagues who consults for Children's Healthcare of Atlanta. Her colleague said that based on the information, we were looking at an in-frame deletion, which would likely result in the milder version of DMD, which is BMD. This news was a gift to us.

Jamie went above and beyond for me and my family by doing so much more than bridging gaps. She was the glue that held it all together — a key figure managing each front as the mediator and liaison for everything. She set our expectations by articulating there would be uncertainty on this journey. She plotted actionable steps to utilize wait times to our advantage and focus on other elements. She explained each complicated step in a way we could understand, provided facts, answered questions, and gave us options. If she didn't have the answers, she connected us to the people who did. As we embarked on each step, Jamie was always waiting in the wings to filter the information and help us pivot if needed. This type of guidance changed everything for us.

Jamie's support and counsel gave us two valuable gifts — perspective and empowerment. Our mentality shifted from something that was happening to us, to one of empowerment. She gave me the confidence and knowledge to make choices for my unique circumstances and the freedom to choose what was best for my family. It felt like she was handing the reins back to us.

Last year, Jamie asked me to speak at the annual National Society of Genetic Counselors conference in a session called "The Changing Landscape of the 'Carrier' Experience." It was a full circle moment because I got to tell her colleagues how she impacted my life. I learned that I could advocate for how important the role of a genetic counselor is and could use my experience with Jamie to pay it forward and help others. In my experience, a genetic counselor can make all the difference. After the conference, I was asked to speak to genetic counseling students in British Columbia and was also contacted by a genetic counselor who had clients who were starting the DMD/BMD journey with their son.

Ultimately, we decided not to pursue IVF for several reasons. The pandemic halted progress during a critical time, since I was approaching 40. Also, Everett was diagnosed with focal cortical dysplasia, a congenital abnormality that can cause seizures, which took on its own journey. My mother and brother also got tested and were found to have the mutation.

Jamie was our beacon of light on a dark path, and I have the deepest appreciation for the unwavering guidance she gave us during our journey.

L TO R: JAMIE, EVERETT, MELEAH

“Jamie went above and beyond for me and my family by doing so much more than bridging gaps. She was the glue that held it all together—a key figure managing each front as the mediator and liaison for everything.”

Brighton Goodhue
Our Anchor

Brighton Goodhue
MS, LCGC

Vanderbilt University
Medical Center
Nashville, Tennessee

Nominated by
Kelsie Hughes

Photographs by
Jackie Osborne

MY HUSBAND, MICHAEL, and I both have a genetic condition called achondroplasia, the most common form of dwarfism. People with achondroplasia are generally healthy and live fulfilling lives. However, if two people with achondroplasia plan to start a family, there's a one in four chance that their baby will be "double dominant," or have homozygous achondroplasia. This means the baby carries two copies of the achondroplasia gene, which is not compatible with life.

We knew this when we became pregnant in November 2020. I'm a pediatric nurse practitioner in oncology, and Michael is a pharmacist, so we both have strong medical backgrounds. When I got a positive pregnancy test, we immediately called the maternal-fetal medicine department at Vanderbilt University Medical Center, where we got a prompt appointment with Brighton.

The moment I met Brighton, I felt at ease. Besides my husband, I've never clicked with someone so easily in my life. When you have a disability, some medical providers talk down to you, but Brighton realized that we were both knowledgeable about achondroplasia and personalized her care to match our needs. A baby born with homozygous achondroplasia will most likely pass away soon after birth. Their lungs are much smaller than the average newborn, and their bodies can't produce enough red blood cells. It's a heartbreaking scenario that Michael and I hoped to avoid.

Brighton knew time was of the essence, so she arranged for me to have chorionic villus sampling (CVS), which determines if a fetus has genetic conditions. Unfortunately, the doctor was unable to get a sample of my baby's placenta due to its difficult location. We then had to wait until we could safely have an amniocentesis in my 16th week of pregnancy.

When you have a disability, some medical providers talk down to you, but Brighton realized that we were both knowledgeable about achondroplasia and personalized her care to match our needs.

The day my amnio was scheduled, there was a freak snowstorm that caused many delays. Brighton called and said, "If it was up to me, I'd walk to your house in the blizzard and do it in your home." Despite this setback, she worked tirelessly behind the scenes to ensure my procedure was not further delayed given its urgency.

We still had to wait two weeks for the results. It's agonizing to wait, knowing there's a 25 percent chance of a lethal diagnosis. I was about 18 weeks pregnant when Brighton called with the devastating news: The baby was double dominant. As a pediatric nurse practitioner, I was familiar with the universal cry mothers make when they learn that their child will pass away. But in this case, the sound was coming from me.

I lost the pregnancy a few weeks later in March 2021. We named our son Patrick James after both of our fathers. During this dark time in our lives, Brighton was a continued source of comfort and care and truly empathized with our situation. She was one of few individuals with whom we were able to be completely vulnerable.

Later that year, after we had time to grieve, Brighton coordinated multiple in vitro fertilization consultations for us. It seemed reasonable, as specialists would be able to genetically test embryos before implanting them. But due to my anatomy, it would have required abdominal surgery to retrieve my eggs, leading us to try one more time on our own.

I got pregnant in March 2022, the exact same week that I'd lost Patrick a year earlier. Brighton was one of the first people I texted. She was ecstatic for us and was able to book our CVS immediately. This time, the procedure was successful. By the end of my first trimester, I knew I was going to have another baby boy, but this time, one we would be able to take home. Brighton was the one to call us with the news. She later told me she'd bought a bottle of Champagne after my CVS because she had this feeling that this pregnancy would be okay. That November, I gave birth to Henry, during the exact same week that we'd conceived Patrick two years earlier.

We wouldn't have been able to get through our delivery day without Brighton. My short stature meant I was at high risk for pregnancy and delivery complications. She recognized the stress and anxiety I was under and offered constant comfort and guidance. Her presence was a reassuring anchor through our whole turbulent journey.

Today, Henry is 7 months old, healthy and thriving. I keep in contact with Brighton and text her pictures of Henry several times a month. I'm active in the dwarfism community, so Brighton asked me to speak to her genetic counseling students at Vanderbilt. It's important they learn how to be compassionate when patients are faced with the possibility of their babies being born with certain conditions.

Over the last three years, Brighton has been with us from our lowest lows to our greatest joys. There are simply not enough words to describe our immense gratitude that she is in our lives.

L TO R: MICHAEL, KELSIE, HENRY, BRIGHTON

DANA

Dana Goodloe
A Silver Lining

Dana Goodloe
MPH, MS, CGC

University of Alabama at Birmingham
Birmingham, Alabama

Nominated by
Meghan Brown

Photographs by
Stacy Allen

THE FIRST MEMORY I HAVE of seeing my son, Walter, is a fond memory but also a terrifying one. It was in 2019 while watching him on my 20-week pregnancy anatomy scan. I noticed during the exam that the technician seemed especially focused on his brain area. Afterwards the physician came in and told us that we would need a targeted ultrasound with maternal-fetal medicine specialists to examine several brain cysts.

I was confused and scared. To make things worse, six weeks was the earliest I could get an ultrasound appointment. That's where Dana stepped in and worked her magic. I knew her professionally because we were colleagues at the Children's Hospital of Alabama. She heard about our troubles trying to schedule a scan and called the maternal-fetal medicine department at the University of Alabama at Birmingham to get us on a cancellation list. A few days later, a spot opened.

When we arrived at our appointment, I was surprised to see that Dana was our genetic counselor. She didn't usually work in the maternal-fetal medicine department but was asked to cover that day for an emergency. Within minutes, I was so grateful that she was there. She was very patient and explained to my then-husband and me exactly what to expect during the ultrasound.

During the scan itself, I knew from the physician's body language that something was wrong. Dana came to the ultrasound room and held my hand when the bad news was delivered. The scan had identified potential defects in Walter's brain, heart, and kidneys. Dana explained our options, which included an amniocentesis to check for genetic conditions such as Down syndrome or trisomy 18.

The one constant in our lives during those weeks was Dana. She went above and beyond to help us understand what we were facing.

Although our amnio revealed no genetic abnormality, we still had to return to the maternal-fetal medicine department every week for the next eight weeks for repeat scans. Every time, we were told something different, depending on which specialist was there to read the ultrasound that day. Sometimes, they would tell us his heart was deformed, and other times they would say it was perfect. It was literally like getting hit by a bus every Tuesday. Just when we would build up some hope, we'd get whammed again.

The one constant in our lives during those weeks was Dana. She went above and beyond to help us understand what we were facing. She connected me to a geneticist who encouraged us to submit Walter's genetic information once he was born. He would be part of a genomic sequencing study that might reveal a gene mutation that could help inform our decisions for his future care.

Walter arrived on July 9, 2019, at 28 weeks. He looked pretty good that first day, and it was clear my little boy was a fighter. But although they tried all kinds of methods and measures to help him, his body was just not compatible with life. After that first 24 hours, he began to deteriorate rapidly and labored to breathe even on a ventilator. On his second day of life at 2:00 p.m., we decided to take him off life support. He died a few hours later. Through it all, Dana was by my side. She was one of the first people to visit him in the neonatal intensive care unit, and she would check in on him — and me — every several hours.

Three months after Walter's death, we received his genetic sequencing. Dana gently sat me down and took me through the report. It turns out my ex-husband and I share a recessive genetic mutation, PPA2, which we both passed on to our son. It's an incredibly rare mutation that was only identified in 2016. There are less than 30 cases of people born with this mutation in the world. My baby was one of them. Most children with this mutation die in infancy, and virtually none make it into adulthood.

It's been almost four years since Walter's death, and I still talk to Dana at least once a month. I'm proud to consider her a close friend. We have a lot in common, as we are around the same age and even attended the same college. She is the one silver lining to have come out of this heartbreaking process. But even before then, I witnessed her fabulous work as a genetic counselor, and I know she gives the same care and attention to all her patients. I'm beyond grateful for Dana and the amazing work she does every day.

L TO R: MEGHAN, DANA

KAREN

Karen Huelsman
The Best Present

Karen Huelsman
MS, LGC

TriHealth Precision
Medicine Institute
Cincinnati, Ohio

Nominated by
Mary Orloff

Photographs by
Matthew Allen

Karen Huelsman by Mary Orloff

I HAVE A STRONG FAMILY HISTORY of breast and ovarian cancer. My grandmother, mother, aunt, sister, and cousin have had either breast or ovarian cancer. My grandmother died in her early 60s from ovarian cancer, and my mom passed away from ovarian cancer when she was only 38. She left behind my dad, me (at 16 years old) and my four younger siblings, ages 14, 13, 11, and 10 months. Before my Aunt Judy died, she accompanied my younger sister and me to get genetic testing as part of a research study. The results came back inconclusive.

I met Karen Huelsman in 1999. Little did I know that my family and I were starting a lifelong journey with Karen, and that she would be guiding us with her knowledge and compassion the entire way. After my initial meeting with Karen, I had genetic testing that showed I was BRCA1 positive. I wasn't surprised by the result, but I did feel unsettled. I was a busy mom with four young children and a job as a research assistant at Shriners Hospital for Children. Now, I had a new label: previvor.

After great deliberation, I had prophylactic surgeries to reduce my risk of breast and ovarian cancers. After that, I started volunteering with FORCE (Facing Our Risk of Cancer Empowered). Karen attended events with me over the years. We participated in panel discussions, lectures, conferences, and support group meetings to educate people on hereditary cancers. Karen is always willing to lend a helping hand and to offer her expertise in genetics. She is a great role model, and I wanted to help women make difficult healthcare decisions, just as Karen helped me.

Her knowledge, compassion, and caring attitude are just a few aspects that make her such a wonderful genetic counselor.

We crossed paths again in 2019, after my husband, Darren, was diagnosed with prostate cancer. I called Karen because Darren's father had prostate cancer, and we were wondering if there was a genetic connection. Although the tests revealed no hereditary link, that knowledge was important for us to share with our family. After years of treatments, Darren is currently cancer free.

Then in 2021, 22 years after I first met Karen, I found myself in another genetic counseling session with her, but this time it was with my four sons, a daughter-in-law, and Darren. Everyone was in town for my son's wedding celebration. With grandchildren on the horizon, we were thinking about the next generation, and we reached out to Karen. Karen was able to coordinate a family session for us on updated BRCA1 information and genetic testing. It was the day after Thanksgiving, a day I knew Karen took off, but she came in with an assistant to provide my family with counseling and support. My four sons decided to get tested that day, and Karen arranged for their blood work.

On December 13, Darren and I were strolling through the Cincinnati Zoo's Festival of Lights when our phones started ringing. First our son Kevin called, then Kyle, Keith, and finally Kenny. The news they shared was nothing short of a miracle — they all tested negative! There was a 6.25 percent chance of those results occurring. I immediately called Karen and told her she just delivered the best Christmas present ever. I think Karen was just as happy as we were, knowing that our sons and grandchildren would not have to think about cancer risk in the same way I had to.

Karen is an expert in the field and is able to explain complicated concepts in an easy, accessible manner. Her knowledge, compassion, and caring attitude are just a few aspects that make her such a wonderful genetic counselor. Last year, I heard that Karen took another job in a genomics lab. I was happy for her, but sad for me and all the other patients she helped over the years at TriHealth. When I recently heard that Karen returned to TriHealth because she missed working with patients, I wasn't surprised. That is where her heart is, and why she deserves the Heart of Genetic Counseling Award.

L TO R: KAREN, MARY

DAWN

Dawn Laney
Above and Beyond

Dawn Laney
MS, CGC, CCRC

Emory University School of Medicine
Atlanta, Georgia
ThinkGenetic Co-Founder

Nominated by
David Jacob

Photographs by
Anna Kariel

I HAVE HEARD that for rare genetic conditions, it takes an average of eight years to get a diagnosis. But in my case, I had to make my own genetic counselor and wait 40 years. That may sound puzzling, but let me explain.

I was born with a congenital heart defect and had open heart surgery at age 5 to repair my heart. My little sister, Carol, was born with the same condition and also had surgery at age 5. I grew up normally and assumed whatever I had was fixed. I eventually became a professional bowler, Navy pilot, and then went into the technology business.

My daughter, Dawn, has always been fascinated with genetics. She got a master's degree in medical genetics and became a certified genetic counselor. While I was building my businesses, Dawn grew her practice at Emory University, where she now is an associate professor and director of the Emory Genetic Trial Center. Her specialty is lysosomal storage diseases, particularly Fabry disease.

In 2007, I got double pneumonia while we were visiting Dawn when she was pregnant with her first son. During treatment, I had a chest X-ray, which showed a large, aortic aneurysm. I called Carol and told her to get an echocardiogram, which showed a smaller aortic aneurysm. At that point, Dawn suspected our diagnoses may have a genetic connection.

The next Christmas, Dawn gave us genome sequencing as our gifts. So, our family gathered around the Christmas tree, and she drew our blood samples.

My results showed that I had a connective tissue disorder, but not a known condition. Dawn sent me to see Dr. Hal Dietz, an expert in connective tissue disorders at Johns Hopkins. Dr. Dietz confirmed that I had familial aortic disease, but he didn't have a specific diagnosis. We would have to wait for more research to occur before we could find an answer.

In counseling patients, her goal is to educate, support, and empower patients to advocate for themselves and their families, so they can live their best lives possible.

That answer came three years later when, after re-evaluating my sequencing, I was diagnosed with cutis laxa — a rare, inherited, connective tissue disorder that is characterized by loose hanging skin. I've always had loose skin, and my mother used to say I would grow into my suit, which I never did. Like she does for all her patients, Dawn got involved and identified a cutis laxa clinical trial, which I enrolled in, and also found me a cutis laxa support group.

Since it took me 60 years to figure out my diagnosis, I wanted to help other people with rare and genetic conditions shorten their diagnostic journeys. I spoke with my business partner, Len Barker, and Dawn about starting a company called ThinkGenetic. In my research, I found there are over 400 million people in the world living with one of 7,000 rare diseases, and only about 25 percent of them get diagnosed.

We wanted to find a way for people to get diagnoses more quickly so they can get the treatment they need sooner. Dawn coordinated over 100 genetic counselors to help us build content for a system that uses algorithms to help people identify if they have a genetic condition. This system can also analyze healthcare records to help hospitals identify patients with rare conditions who might have been missed.

But that's not all Dawn has accomplished. She is considered an expert on Fabry disease and speaks at conferences around the world about her research. She meets with patients and families and hosts patient advocacy meetings. In counseling patients, her goal is to educate, support, and empower patients to advocate for themselves and their families, so they can live their best lives possible.

She also volunteers at the Fabry Family Weekend Camp every year. The camp is a great way for people to connect — through educational sessions for the parents, and also so kids can play with their peers. I volunteer at the camp and have heard from patients how much Dawn helps their families and how they consider her not just a genetic counselor, but a friend.

Dawn has also written several children's books on rare diseases to help patients and families understand their conditions, as well as a workbook to help parents manage Fabry disease and their families. One of her patients, who has Fabry and is great with graphics, illustrated her children's books.

In 2016, the National Organization for Rare Disorders recognized Dawn with the Rare Impact Award for her work on the disease and with the Fabry community. More recently, Dawn has been advocating for newborn screening in the state of Georgia, which would help uncover genetic conditions earlier.

So, not only did I create my own genetic counselor, but I made one who assisted my sister and me in finding our diagnoses, who helps countless families and patients navigate rare diseases, who dedicates her life to finding cures for some of the most difficult conditions, and who is educating generations of genetic counselors at Emory University. I am so very proud of her!

L TO R: DAWN, DAVID

MANDY

Mandy Miller
Winning the Lottery

Mandy Miller
MS, CGC

Indiana University School
of Medicine/Department of
Medical and Molecular Genetics
Indianapolis, Indiana

Nominated by
Valerie Stinger

Photographs by
Lauren Ballenger and
Angela DeCenzo

Mandy Miller by Valerie Stinger

AT THE END OF MARCH, I felt I'd won the lottery — but not for the reasons you might think. A decade ago, I was diagnosed with Parkinson's disease. I've been able to manage my symptoms and maintain a vibrant lifestyle thanks to the efforts of my wonderful medical team here in Palo Alto, California. However, that would have been thrown in jeopardy if it weren't for the intervention of Mandy, a genetic counselor at the Indiana University School of Medicine. She may have preserved my quality of life.

My story with Mandy begins in March. I was connected to her through my local Parkinson's support group. Our moderator sent an email to the group about the Parkinson's Foundation PD GENEration project at the Indiana University School of Medicine. They were looking for volunteers to undergo genetic testing. I decided to be a good corporate citizen and sign up. As part of the study, my sample would be part of a large genetic database. I didn't expect to learn much that would be relevant to my current treatment, but I liked the idea of furthering Parkinson's research.

The Foundation sent a kit, I did a cheek swab, and sent it in for analysis. I heard from Mandy on March 30. I was struck by how professional and thorough she was. Mandy reassured me that I didn't need to take notes — she wanted me to focus on listening, and that she would send a thorough report later. She took 45 minutes to go through the results and explain the implications. It turns out that I was one of the 5 to 15 percent of Parkinson's disease patients who have mutations in the GBA gene, which is the most important genetic risk factor for the condition. During our conversation, I mentioned that I was preparing for a surgical procedure known as deep brain stimulation (DBS). This treatment is often used to help relieve motor symptoms of Parkinson's, such as tremors and stiffness.

That's when everything changed. Mandy told me that my genetic profile was associated with an increased risk of cognitive decline. She emailed me the report immediately so that I could show it to my surgeon. One of the possible complications of DBS is cognitive decline, and while the risk is low across the whole population of DBS surgeries, she worried that my gene variant put me in a much more vulnerable subset.

It turns out she was right on the mark. Recent research shows that Parkinson's disease patients with the GBA gene mutation who undergo the type of deep brain stimulation that I was set to receive are much more likely to experience cognitive decline. As soon as my surgeon and neurologist learned that I had the mutation, they agreed that the surgery should be indefinitely postponed. The risk was just too great.

When I heard that, I felt like I had won the lottery. Yes, it was disappointing not to be able to get the treatment, but I dodged a bullet. As my neurologist told me, if we hadn't known about the mutation, I could have gone ahead with DBS and might have been fine. Or I could have had a negative outcome, and we wouldn't have understood why. I would have ended up as just another statistic. I'll never forget my doctor saying, "It would be unconscionable for us to proceed with putting you at that level of risk."

Life with Parkinson's disease is no picnic. I'm on several different medications to help manage motor symptoms, which include stiffness, tremors, and muscle cramps. Sometimes it seems like I spend all my time counting pills and exercising (I do a lot of walking, yoga, and stretching, to help me move around more easily). My physical function has declined already because of Parkinson's disease. If I'd experienced cognitive decline, too, that would have taken away all that I have left.

Thanks to Mandy stepping in, I can continue to live the life that I love. What impressed me the most throughout this situation was her calmness and professionalism. She was never alarmist, and she never told me what to do. She just wanted to make sure I had the information I needed to make the right decision for me.

I'll always be grateful to her and to the team of medical professionals she works with at the PD GENEration project. What they're doing, day in and day out in the name of Parkinson's disease research, is so important and makes a difference in our lives. For me genetic testing wasn't hypothetical or academic — it had real patient management implications. As the criteria for DBS are revised to incorporate the recent findings, genetic testing will benefit many others, and we will all be better as a result of the daily work done in genetic testing labs.

VALERIE

“Thanks to Mandy stepping in,
I can continue to live the life
that I love.”

KATE

Kate Murphy Orland
MVP of My Team

Kate Murphy Orland
MS, LCGC

University of Wisconsin-Madison
Medical Center
Madison, Wisconsin

Nominated by
Laura Woodberry

Photographs by
Narayan Mahon

MY FATHER, UNCLE, AND PATERNAL GRANDFATHER all died of sudden cardiac arrest by the age of 45. I began showing signs of cardiac symptoms in childhood. You would think with my family history that my doctors would have taken seriously my complaints of leg weakness, shortness of breath, and heart palpitations. Instead, I was told that I was overweight and lazy.

Over the years, my symptoms worsened. In my early 20s, I was chasing around two toddlers and going to nursing school full time. I didn't have time to worry that, every now and then, I couldn't breathe or that my heart was pounding so hard it felt like it would explode. When I mentioned it to my primary care physician, he told me it was just anxiety. Then my younger brother was diagnosed with sudden onset heart failure at age 36. His doctors took this seriously and did genetic testing for him. His tests revealed four genetic defects, including the LMNA mutation, which causes cardiomyopathy and more serious conditions, such as muscular dystrophy, sudden cardiac arrest, or heart failure. Subsequent testing revealed that I carried this mutation too.

When I first got the news, I felt validated. I had been telling my physicians something wasn't right for 20 years and was dismissed as a hypochondriac. However, I was also quite scared. That's when Kate stepped in to offer comfort and reassurance.

I met Kate at my very first appointment with the electrophysiologist, a cardiologist who diagnoses and treats issues with the heart's electrical system. She gave me information about my defect, its hereditary nature, and what to expect, and spent over an hour with me answering questions and calming my fears. I was 41 at the time, and all I could think about was that my father passed away at 44, so I only had three years left. I'll never forget the compassion in her voice when she said, "Your dad's history is not your reality. We never know how this condition will progress." She also pointed out that the mutation tends to present less severely in women.

Kate's the go-between, the one who sits down with patients and breaks everything down. She is a most valued member of my healthcare team.

Kate was also there for me when I learned that I passed the gene mutation on to both of my sons. I was frightened and felt incredibly guilty, especially since I knew that men have a worse prognosis. My oldest son began to display symptoms at age 8. His pediatrician diagnosed him with another heart defect a couple years later — a bicuspid aortic valve, which means the aortic valve contains only two flaps, instead of three. Overtime, this can make the heart work harder and potentially lead to heart failure.

Even though my boys tested positive when they were teenagers, they weren't concerned until they were in their 20s, when they wanted to know how the mutation affected overall health and whether they could pass it to their own children. When my oldest son got married, he saw Kate, and she explained that the LMNA mutation was autosomal dominant, meaning there was a 50 percent chance his offspring would have it. As a nurse myself, I could have had that conversation with him, but he needed to hear it from an impartial professional. Thankfully, his beautiful little girl does not have the LMNA gene mutation.

Today, a decade after my diagnosis, I'm in heart failure. I'm constantly short of breath, fatigued, and dependent on a pacemaker as well as a defibrillator. I had to stop working in 2018, since I could no longer tolerate 12-hour days on my feet. It devastated me because I loved being a nurse.

Kate has been the one constant who has always been there for me when I get confusing test results. For example, an ultrasound indicated that my heart was enlarged and then another six months later looked fine. Kate explained that each ultrasound is a snapshot in time. Your heart can remodel. Since I'm on many medications and have a pacemaker, my heart will have periods of time where it will improve, then regress. Whenever my head starts to spin, I can call Kate. She answers all my questions and brings me back to reality.

Case in point: Recently, I switched from a traditional pacemaker to one with cardiac resynchronization therapy (CRT). Unlike other pacemakers that have one or two wires, CRT has three. This makes it easier for your ventricles to work together. Kate took the time to explain in detail what CRT was and why it was a better option for me. Whatever my question, Kate always has the answer.

As humans, it's normal for us to fear what we don't know or understand. Not all healthcare providers get that, and sometimes they don't do a great job explaining. Kate's the go-between, the one who sits down with patients and breaks everything down. She is a most valued member of my healthcare team.

L TO R: LAURA, KATE

Mitchel Pariani
Always By Our Side

Mitchel Pariani
MS, CGC

Stanford University School of Medicine
Stanford, California

Nominated by
Ramona Bywater

Photographs by
Angela DeCenzo and
Ian Spanier

MY SON, QUADE, was diagnosed with Marfan syndrome 18 years ago at the age of 6. Marfan syndrome is a rare, genetic condition that affects the connective tissues of the body. Over time, it can damage blood vessels, skin, lungs, eyes, certain bones, and the heart. In the heart if the aorta weakens, it can balloon out and break, which is life-threatening. As a result, Quade has been monitored very carefully.

When Quade was 10, we heard about a study for Marfan syndrome at Cedars-Sinai hospital in Los Angeles. At that time, people with Marfan syndrome were often treated with atenolol, a beta blocker that is used to treat high blood pressure. The study at Cedars-Sinai wanted to see if another common blood pressure medication, called losartan, would be just as effective in kids and young adults. Quade was doing very well, but our doctors did warn us that his aorta was weakening. In our minds, we had nothing to lose if we enrolled him in the clinical trial. Quade wanted to do it as well — he liked the idea of giving back to his community.

When we arrived at Cedars-Sinai on a sunny July day in 2009, Mitchel was the first person we saw. His smiling face put us at ease. He walked us through the entire facility and labs so Quade could see everything, including the laboratory where he would get his blood work and the imaging center where he would get X-rays and echocardiograms of his heart. When you have a young child with a serious medical condition, you can feel overwhelmed and alone. Mitchel was always there by our side, supporting us along the way.

The first time Quade had to have blood work done there, he had a very difficult time. The technicians couldn't get to the vein,

When you have a young child with a serious medical condition, you can feel overwhelmed and alone. Mitchel was always there by our side, supporting us along the way.

and it was very painful. Mitchel checked in on us and suggested we go to lunch and try again. The second time worked like magic, and we were able to get the tests we needed done. We also lived 90 minutes away from the hospital. Mitchel would always schedule all our appointments together, so we could do everything — lab work, imaging, and doctor appointments — all at once. It made our lives much easier.

We saw Mitchel every six months like clockwork for three years. After the trial ended, we learned that Quade had been randomized to get losartan. (The study found that patients given either medication did equally well.) Today, he is on both losartan and atenolol and is doing great. Since our clinical trial, other research has shown that this combination is the most effective drug regimen to prevent aortic enlargement.

Once the study ended, we still saw Mitchel every year at annual Marfan syndrome conferences. A few years ago, Mitchel left Cedars-Sinai and took a job as a genetic counselor at Stanford.

When Quade's local doctors recommended that we form a relationship with a heart surgeon for possible future use, Mitchel gave us a wonderful recommendation for a surgeon at Stanford, if we wanted to make the trip. Thankfully, Quade's aorta eventually stabilized, so he didn't need the surgery.

Quade's now 24 and doing great. He is a full-time professional and is very active in Marfan syndrome fundraising and advocacy. He keeps in touch with Mitchel too. For us, Mitchel is more than a genetic counselor — he's like family.

RAMONA

Nicole Pederson
One of the Family

Nicole Pederson
MS, CGC

Essentia Health-Duluth Clinic
Duluth, Minnesota

Nominated by
Laura Perttula

Photographs by
David Ellis

I COME FROM a very large family. My grandpa had seven siblings, my mother had 13, and I am one of 37 grandchildren. There are hundreds of us, so when a lethal genetic mutation surfaced in our extended family, the work required to explore our family health history and coordinate genetic testing was as big as our family. Handling just us would be a full-time job, and more if you have other patients to see. Thankfully, Nicole Pederson was up to the task.

In January 2023, my cousin forwarded me a letter informing us that a cardiac gene mutation, PKP2, had been found in our family. PKP2 is associated with a disease called arrhythmogenic right ventricular cardiomyopathy (ARVC), in which fatty fibrous tissue replaces normal heart muscle. As this fibrous tissue builds up over time, it can cause abnormal heart rhythms or even sudden death. Activities that raise your heartrate, like running, can increase that fatty tissue buildup more quickly.

This news was upsetting to me, because I have been a runner for 10 years, as well as an occasional triathlete. The possibility that what I had considered a healthy activity could be deadly was ironic. Running is also how I deal with stress, so having that outlet taken away was particularly distressing. Although this variant primarily impacts youth athletes, I didn't start getting active until 10 years ago.

So far, Nicole and her partner have tested close to 100 of our family members. Because of our family's size and our close geographic proximity to each other, Nicole has even helped launch a clinical trial based on our family's testing.

I was the first in our family line to meet with Nicole. I was nervous, because one of the potential consequences of having the mutation could be deadly — one of our relatives had died very young while playing at a high school football game. At the time, they didn't know he had the mutation, but because they tested some of his preserved tissue, we found out that he had the mutation. Other people in the family have the mutation and had surgery, and yet others have been asymptomatic.

At our meeting, Nicole was warm, compassionate, and very reassuring. She's also very friendly — you would want to have coffee or a drink with her. She could joke around with me but in a way that was still respectful and put me at ease when I was anxious.

Nicole explained the mutation to me and the testing process, she walked me to the lab, and then met with me after my test results came back to discuss next steps. She also met with my sister, who has four children and six grandchildren (including a 15-year-old hockey player who would be at a high risk, if he had the variant), along with numerous other cousins. Like she did with me, Nicole guided them through this process, reassuring them they would get through this.

So far, Nicole and her partner have tested close to 100 of our family members. Because of our family's size and our close geographic proximity to each other, Nicole has even helped launch a clinical trial based on our family's testing. If you have the mutation, the chances of passing it down to your offspring are 50 percent. As more of us go in for testing, some of my cousins and I have joked that we feel like we're playing the genetic lottery.

In the end, I did not win the genetic lottery and tested positive for PKP2. Nicole immediately stepped in and recommended that my primary care physician refer me to a cardiologist, who benched me from running and most of my other workouts until I had extensive testing. During those two months, I had an electrocardiogram (EKG), an echocardiogram, a cardiac MRI, and a Holter monitor, all of which revealed that I was asymptomatic, which is great news. While I'll continue to have a yearly EKG and check-ins with my cardiologist, I have been cleared to start running and resume my other normal activities again.

Once a month, we have a family happy hour. As I mentioned, Nicole is someone you would want to have a drink with, and I've invited her to come. I found out later that other family members, including my 71-year-old cousin, a retired fire chief — a tough guy — had done the same. She is just lovely and fits right in with all of our various personalities. She could truly be a member of our family.

L TO R: LAURA, NICOLE

SARAH

Sarah Saxton
Our Unexpected Ally

Sarah Saxton
MS, CGC

Molecular Genetics Laboratory,
NYC Office of Chief Medical
Examiner
New York, New York

Nominated by
Eric Wahl

Photographs by
Benjamin Norman and
Tegra Stone Nuess

WHEN MY BROTHER, Grant Wahl, died suddenly and unexpectedly while covering the World Cup in Qatar this past December, my family was faced with a wave of unenviable tasks amidst our shock and grief. My brother was a public figure — a sports journalist with an international reputation. My family and I had to find ways to juggle our grief and astonishment at the loss of Grant with the public, understandably, wanting information about his death. It was an extremely difficult time for all of us.

After Grant's body was flown to New York City for an official autopsy, the New York City Medical Examiner's Office, which is the only medical examiner's office in the United States with a postmortem molecular diagnostics lab, discovered Grant's cause of death — an aortic aneurysm — as well as a genetic mutation on the 15th chromosome referred to as FBN1. Unsurprisingly, this new knowledge prompted concern in our family — was this genetic condition present in other family members? What should we do next? How would we go about marshaling the necessary information and providing it to the right people?

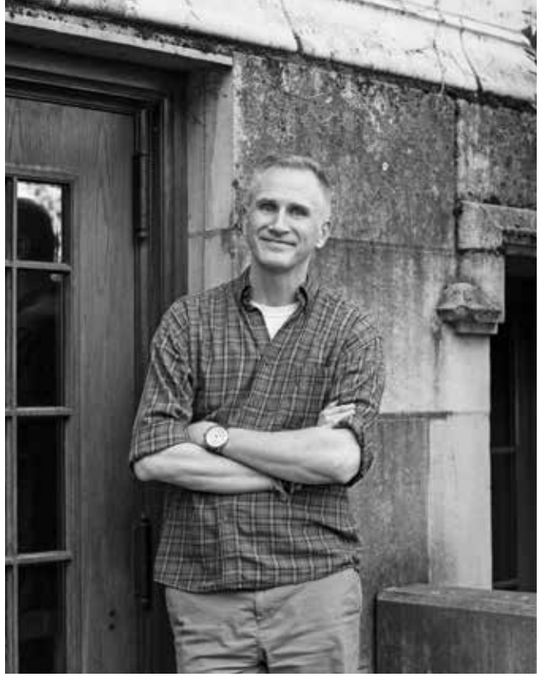
Less than a month after Grant's death, Sarah Saxton phoned me to begin a series of conversations about the FBN1 mutation. I was shell-shocked at the time and in the midst of profound grief, but she helped me to feel less scared and to understand just how unique a situation ours is. On that first call, Sarah told me what is known about the mutation and how researchers are continuing to study its characteristics and origins to help others in the future. Her kindness, calmness, and thoughtful responses to my questions and concerns set me at ease and spurred me on to learn more. In addition to discussing my next steps and how to go about them, she made a point of asking how I was feeling and was so understanding when I told her. I knew then that I had an ally and was better equipped mentally and emotionally to go forward with testing.

Part of what I respect about Sarah is not only her knowledge, but also that she knows how to use that knowledge to help people. Coming from a teaching background, I know this is a real endeavor and that it takes practice, humility, and a kind of understanding of people that not all medical providers are required to have.

As a teacher and a professor, I have dealt with students in crisis situations, but I can't imagine that level of support being a regular component of my job. In my 53 years, I have never met anyone like Sarah. I was overwhelmed with respect for her ability to have these kinds of conversations with people like me who have just suffered a devastating loss. I now know she is the only genetic counselor in the United States dedicated to helping families who have lost loved ones.

Before my experience with Sarah, I thought the phrase "genetic counseling" related to family planning, but now I understand the tremendous impact it can have for all people. It's also easy to imagine laboratories as being divorced from emotional components. Part of what I respect about Sarah is not only her knowledge, but also that she knows how to use that knowledge to help people. Coming from a teaching background, I know this is a real endeavor and that it takes practice, humility, and a kind of understanding of people that not all medical providers are required to have.

After undergoing genetic testing, I learned that I share the same FBN1 mutation as Grant, and with Sarah's help, I have been able to receive the kind of medical testing and care that might have saved my brother had we known about his condition.



ERIC

Every year for the rest of my life I will have heart scans, and I know what kinds of exercises I can and cannot do.

I have come to realize and appreciate that Grant's death may have saved my life and the lives of our family members who share the same genetic mutation. In this way, I can honestly say, without hyperbole, that I feel as though I also owe my life to Sarah Saxton and the work of her team at the New York City Office of Chief Medical Examiner. I want people to know that this work is being done. With Sarah's guidance, we opened our family up to further study, which means we can help more people. I know that would make Grant proud.

MICHELLE

Michelle Springer
The Gift of Possibilities

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MICHELLE SPRINGER WAS THE FIRST person to help me realize I was not a ticking time bomb. My mom died of breast cancer at 35 when I was only 6 years old. I grew up with my dad — a superhero — a single parent who raised three kids on one income. From a young age, I always assumed I would get diagnosed, and for years, doctors urged me to get genetic testing.

When I was 16, my doctor found lumps in my breast. Although they were benign, their presence was a stark reminder that it was just a matter of time before I developed cancer. While my doctors kept recommending testing, I distinctly remember why this was such a terrifying proposition. My primary care physician had said to me, "If you get genetic testing and you test positive, you just have a double mastectomy and move on."

I was 16. I didn't even know what a double mastectomy was, let alone what the ramifications of having one would be. At the time I thought, if that's my only option, I'm not getting tested. I didn't feel like I had choices.

In my early 20s, my family health history kept surfacing at every doctor's appointment. Coincidentally, a close family friend of ours was the charge nurse at the hospital's breast center. She recommended that I talk to this amazing woman she worked with — a genetic counselor named Michelle.

Even though I had been encouraged for years to pursue genetic testing, I had never been offered genetic counseling. I didn't know what it was, but I was intrigued. After our friend reassured me that I did not have to undergo genetic testing if I spoke with Michelle, we set up a time to talk over the phone. On that phone call, Michelle told me everything that I had needed to know all of those years ago.

She told me what to expect at our appointment — what it would mean if I tested, what it would mean if I didn't, what it would mean if I tested positive, and what it would mean if I tested negative. She explained how we would look at my family history and how she would unpack all the science behind it if I wanted. At the end of the appointment, she said it would be my choice to get tested, and if I decided not to, she would discuss other options, such as screening.

Options. This was the first time I ever had options in my life. Until now, every doctor had approached me as if I was on the same path as my mom.

I knew then that I wanted genetic testing, because Michelle made me feel like a positive result wasn't a death sentence or wouldn't require an immediate surgery. When I did test positive for the BRCA mutation, Michelle, as promised, gave me options. For a while, I chose screening every six months, and then, I decided to have a double mastectomy. All of it was my choice.

Michelle put me in touch with a support group specifically for young women with a high risk of breast cancer, a group that became a huge support for me. She also connected me with other women who were fearful of being tested, as I had been. I found it helpful and empowering to share my story, my knowledge about testing, my options, and even the scary parts that I knew they were experiencing too. I was flattered that Michelle thought of me to give that support to other women.

I'm now 35, and I have known Michelle for more than 10 years. She continues to have a profound impact on my life and that of my family. She has helped my dad and maternal aunt get tested — he is a carrier for the BRCA mutation, while my aunt is negative. Because I was initially tested so long ago, Michelle helped me get retested in case I had markers or mutations more recently identified. I have even joined her on occasion to discuss the importance of bedside manner when she gives lectures on genetic counseling to medical students.

Michelle has done so much for me, but most of all, at a time when I felt like I had no options, Michelle gave me a world of possibilities — and for that I am forever grateful.

CALEY

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The Future of Genetic Counseling in a Post-Affirmative Action United States

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Photograph by
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I AM WRITING THIS ESSAY in the aftermath of three U.S. Supreme Court decisions that strike down affirmative action, open the door to discrimination against protected classes of people, and bar President Joe Biden's student loan forgiveness program. These rulings come at a precarious time in our nation's history and affect the future of genetic counseling in many ways.

A concerning pattern of civil rights victories that survive less than one human lifetime exists in the U.S. Reconstruction, Roe v. Wade, the core provisions of the Voting Rights Act, and now affirmative action have not lasted. The forces that aligned against affirmative action are not resting on this victory and are already challenging scholarships designated for students of color. At the time of this writing, they have had at least one win. This pattern does not bode well for the Americans with Disabilities Act, marriage equality, and equal employment opportunities.

The Supreme Court decisions will have a cumulative impact on health equity in our country. Income inequality in the U.S. has been expanding for decades and has continued to escalate. Lower rates of undergraduate, graduate, and professional school attendance would lead to a loss of generational wealth in marginalized communities. Loss of abortion access in socioeconomically disadvantaged communities may also reduce enrollment in higher education. Health inequities would deepen due to increased poverty and decreased diversity in healthcare professions.

The genetic counseling workforce has been diversifying, as reflected in admissions match statistics. Since 2018, students of color have made up over 30 percent of incoming cohorts, a sea change from less than 10 percent in the past. However, Black, Indigenous, and Latino students are still significantly underrepresented, and increasing their numbers will now be more challenging. Affirmative action bans in several states have reduced the percentage of underrepresented students in higher education, particularly in STEM fields and healthcare professions. In the coming years, there will be fewer prospective students of color to recruit into genetic counseling programs, and other programs and professions will also compete for these students.

My hope is that by the time this essay is published, we are developing new and sustainable systems and investing in existing programs to continue diversifying our workforce in all dimensions of identity so that we can better serve patients from all walks of life.

The students of color who can navigate this newly narrowed gauntlet will have to be the cream of the crop, and we need to redouble our efforts to attract their attention. Many students of color feel pressure to make career decisions before or early in college to assure they will get good jobs upon graduation, so we need to reach them before college.

Different methods have been utilized by genetic counseling programs to attract diverse candidates and it is critical to reinforce these initiatives. Holistic admissions and optional GRE scores have helped to increase the diversity of some programs, but not all programs have the independence within their universities to make such choices. Universities typically have programs for first-generation undergraduate students, but these are rare at the graduate level. Genetic counseling programs could advocate for services for first-generation graduate students through existing programs for undergraduates or through collaborations with other graduate programs. This would attract a more diverse applicant pool.

Seeing people of color in leadership positions makes an indelible impression on prospective students. The hiring of diverse faculty has expanded in recent years and will appeal to diverse applicants. A mentoring pipeline to prepare these faculty members for program leadership roles will be crucial as the number of genetic counseling programs continues to grow.

Genetic counseling programs need to ethically recruit prospective students by providing more information on financial aid and the true cost of attendance to prospective students so that they can make informed decisions about where to apply. The hidden costs of attending genetic counseling school can include housing, transportation to rotations, and attendance at the National Society of Genetic Counselors annual conference. Currently, there are students relying on food assistance and some considering leaving school. Some students must work, which can impact their academic performance. These issues do not only affect students of color.

My hope is that by the time this essay is published, we are developing new and sustainable systems and investing in existing programs to continue diversifying our workforce in all dimensions of identity so that we can better serve patients from all walks of life. It is imperative that we do so in the face of the deepening inequities in our country. We must be vigilant to protect any victories, since the lessons of history show us that the work of justice never ends.