

## 2012 National Society of Genetic Counselors Presidential Address: Maintaining Our Professional Identity in an Ever-Expanding Genetics Universe

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Good morning, everyone. I am thrilled to be here and honored to be selected as your 2012 NSGC president. I would first like to thank and acknowledge Karin Dent, outgoing President, and Meghan Carey, NSGC's Executive Director, without whom I would never have felt prepared to take on this daunting role.

I'm going to talk today about how I've seen our profession grow over the past 26 years that I've been in practice. My career has veered off in a direction that one would not traditionally think of as genetic counseling, and yet I still maintain my identity as a genetic counselor. When I graduated from the Sarah Lawrence College program in 1985, there were only two career options for graduates: prenatal and pediatric genetic counseling. Yet look at where we are today! In the ever-expanding universe of genetics, our field has branched out into many different areas of professional practice, including cancer genetics, personalized medicine, fetal therapy, education, research, and pharmacogenomics. As our professional diversity has grown, we've come to talk about traditional and nontraditional roles, but I contend that this distinction is no longer valid. We have all branched out in terms of expertise, so that even in areas like prenatal counseling, a specialty at the very core of the genetic counseling profession, there continue to be new developments and new service delivery models that keep it relevant

and moving forward. Therefore, in my first presidential decree (and I believe I can make presidential decrees starting January 1st!), I propose that we retire this outdated notion of traditional versus nontraditional genetic counseling.

I'd like to tell you a bit about where my own career path has taken me. Upon graduation, I took a job at Elwyn, a nonprofit human services organization that has been around since 1852. Elwyn is the oldest continuously operating agency of its kind in the world, an organization steeped in history and very much at the center of the intellectual disability field. In addition to affiliated sites across the United States, Elwyn's main campus is located outside of Philadelphia on 500 rolling acres where we run a number of innovative programs for children and adults with special needs. Elwyn is neither a medical center nor a university, so right out of the gate, I knew I'd started down an atypical career path. I found myself on a campus with 1,100 employees, among whom I was the only one with expertise in medical genetics. I was one of a small handful of health care professionals at Elwyn, my other colleagues being special educators, psychologists, and nonmedical staff involved in the education and support of children and adults with intellectual disabilities.

When I was hired, my initial charge was to work with our small medical team to coordinate diagnostic genetic testing for the hundreds of individuals and families accessing Elwyn services. Because those services run the gamut from early intervention to geriatric care, I found myself interacting with infants, senior citizens, and everyone in between. In 1985, there was a relatively good cytogenetic test for fragile X syndrome, and because it's a common cause of intellectual disabilities, we were quickly able to diagnose over 100 families with fragile X within my first few years at Elwyn. Serendipitously, just as we were finding large numbers of

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people with fragile X syndrome, knowledge about this disorder was also expanding. Particularly after the identification of the *FMR1* gene in 1991, new discoveries were being made about the effects of pre- and full mutations on females, about the association of fragile X with autism, and about the neurocognitive profile of people with the syndrome. As this information was emerging, I found myself at ground zero of the intellectual disability world, surrounded by special education and psychology professionals, few of whom had ever heard of fragile X or other genetic disorders besides Down syndrome. For me, the idea that genetic syndromes could be associated with specific behavioral phenotypes and neurocognitive profiles was particularly intriguing, and this soon became a passion and the focus of my work. A student with Prader-Willi syndrome, for example, could have a relative strength in visuospatial processing, while his classmate with the 22q11.2 deletion syndrome had severe visuospatial deficits, and yet they both had the same IQ. These findings had important practical implications for intervention; however, special education had historically paid little attention to etiology and was essentially using a one-size-fits-all approach. While the notion that “etiology matters” is not at all surprising to genetic counselors, in the intellectual disability world where I worked, this concept was both new and controversial.

Over time, I began to hone expertise in several different genetic syndromes, always with particular attention to cognitive and behavioral phenotypes. Early on, I became interested in Smith-Magenis syndrome, and my colleagues and I published research on self-injury, personality, and other behavioral aspects of this disorder. We also developed expertise around the 22q11.2 deletion syndrome and its relationship to psychopathology and certain kinds of unusual learning disabilities. I was spending a lot of time in schools and working mostly with nonmedical colleagues in special education and psychology. After a number of years of going down this psychoeducational path at Elwyn, I would think to myself, “Am I still a genetic counselor?” I often wondered whether my behavioral interests had taken me too far afield from “traditional” genetic counseling.

As my expertise grew, I began to get requests for consultation from schools and agencies throughout the country. A special education director in Iowa would call, for example, and say, “We’re working with a student who has Smith-Magenis syndrome. Can you please come out to educate our staff about this disorder and help us write a behavior plan that best addresses the child’s needs?” Requests for “technical assistance” began coming in from all over the country, and before I knew it, I was running an active consulting business out of the genetics department at Elwyn. I sometimes provided these consultations alone, but more often, I was accompanied by my colleague, Barbara Haas-Givler, who has over 30 years of experience in special education

and is also a board certified behavior analyst. I found myself spending more time in my car than in the office, and at times it felt as though the car *was* my office. There is a lot of talk these days about service delivery models for genetic counseling, and the closest I can come to describing this particular service delivery model is to liken it to the television show, “SuperNanny®”. For those not familiar with the show, it involves a British woman who travels around in her car to see families with unruly children. Over the course of a few days, she helps the family develop and implement a behavior plan. In a parallel way, using our expertise in behavioral genetics, and based on a foundation of published research and a lot of hands-on experience with different genetic conditions, Barbara and I became the behavioral genetics “supernannies”. Week after week, we would go off on syndromic adventures to remote areas — in fact, we were *always* going to remote areas — and Barbara would say, “Aren’t there any kids with syndromes in Las Vegas or San Francisco?” Just last year, we were asked to go to a very rural area of central Canada. It took us hours and hours of driving to get there from the airport. When I tell you it was remote, I can illustrate this by saying that the town itself had fewer than 1,000 people, and the nearest neighboring town was at least an hour and a half away. In the town’s small elementary school, there were only 30 children in the entire student body, ranging from kindergarten through 8th grade. On the morning of the consultation, we were greeted at the school by all 30 students singing the Canadian national anthem. We had tears in our eyes from the beauty of the moment, and Barbara was thinking, “Where *are* we?”, while I was wondering, “Am I still a genetic counselor?” Then we remembered: we came to Canada to see identical twin 7 year olds with Smith-Magenis syndrome, and their family and small community were rallying around them to learn everything they could about the behavioral and cognitive manifestations of the disorder so that they could give them the best possible quality of life.

During those times when I was back at my office, otherwise known as the Elwyn Genetics Billing Department, I found myself pondering and worrying about issues of reimbursement. This is something I share in common with other genetic counselors, but because Elwyn is not a medical center, we do not bill health insurance companies. Instead, genetics consultations are paid on a fee-for-service basis through contracts with government agencies and school districts. Over the years, I have become quite expert at negotiating contracts, understanding developmental disabilities agencies at the state and local levels, and jumping through the many layers of required paperwork to be added to governmental vendor lists. I now know more than I ever wanted to know about filling out W-9 forms and creating invoices. Although it’s not something I would have ever considered a genetic counseling skill, many of us have had

to develop expertise in billing and reimbursement as a necessary part of our professional lives.

Another unanticipated skill that I share with many genetic counselors is event planning. As a board member for the International 22q11.2 Deletion Syndrome Foundation, for example, I worked with families and professionals to plan a major event called “22q at the Zoo” which involved simultaneous awareness celebrations on the same day at 63 zoos around the world in 9 different countries. Mary Delany-Hudzik is a genetic counselor who coordinates the Elwyn Fragile X Center, including its regional parent group; and on any given weekend, Mary can be found bowling with kids who have fragile X, bouncing along on a family hayride, or planning a sibling workshop. Along with my colleague, Barbara Haas-Givler, I’ve worked closely for years with PRISMS, *Parents and Researchers Interested in Smith-Magenis Syndrome*, a family support group which I co-founded in the 1990’s and which has since gone on to become a major international organization. There are hundreds of similar genetic support groups, and behind most of them, there is usually a genetic counselor working alongside patients and families, providing genetics expertise, emotional support, and a fair amount of elbow grease. We should all be very proud of this unique skill set. It’s one that makes a tremendous contribution to the families we serve.

As you can see, my path has taken me in a direction that, prior to my earlier presidential decree, one might have called “nontraditional”, and yet I’ve always identified myself as a genetic counselor. I may have pondered the rhetorical question, but really, I know I’m a genetic counselor and I’ve always been one. (Besides, here I am standing before you as the incoming president of NSGC, so if I’m not a genetic counselor, there is something seriously flawed about our election process!) A few years ago, NSGC convened a task force to identify the key attributes of genetic counselors. There is an excellent document on the NSGC website describing the outcome of this work. To summarize, the task force identified the following core skills of genetic counselors:

- Deep and broad knowledge of genetics
- Ability to tailor, translate and communicate complex information in a simple, relevant way for a broad range of audiences
- Strong interpersonal skills, emotional intelligence, and self-awareness
- Ability to dissect and analyze a complex problem
- Research skills
- In-depth knowledge of healthcare delivery

When I look at these attributes, I can see why so many of us who have gone in different professional directions still maintain our identities as genetic counselors. It’s clear that these core skills represent a perfect starting point for branching out into the whole genetics universe, so it shouldn’t be at

all surprising that our profession has become so diverse in terms of practice models.

I’d like to highlight a few other genetic counselors who have taken their own pioneering paths to move the boundaries of what we consider genetic counseling. Jill Stopfer at the University of Pennsylvania is widely published and well known for her work in cancer genetic counseling. Jill’s a hard-working NSGC volunteer and is the incoming vice chair of our Public Policy Committee. A number of years ago, Jill was one of the first people to utter the words “cancer” and “genetic counseling” in the same sentence. It’s hard to believe that there was a time when the idea of cancer counseling was not a glint in anyone’s eye, and no one considered it a potential direction for our profession. Years ago, I remember hearing Jill talk about this brand new area of cancer genetic counseling, and I was secretly pleased. Like me, she was one of those rogue genetic counselors, out there doing something very different. Since then, cancer counseling has become an integral part of our professional practice, and it all started with thinking beyond defined career boundaries.

Another person who has taken genetic counseling and invested it in a specific area of expertise is Donna McDonald-McGinn, a colleague in my region at the Children’s Hospital of Philadelphia. Donna has become an international expert on the 22q11.2 deletion syndrome. She has worked tirelessly and collaboratively with families, clinicians, and scientists from around the world. They look to her as a leader for her clinical expertise but also for her close engagement with families and dedication to bettering the lives of people with the syndrome. Likewise, Ann C.M. Smith at the National Institutes of Health has dedicated a lifetime of effort toward advancing research on Smith-Magenis syndrome. Many of you who are newer to the profession may not realize that Ann, who was a founding member of NSGC and a past president, is also the “Smith” in Smith-Magenis syndrome. Her interest began in 1982 with a poster at an ASHG meeting in which she reported a patient with a cytogenetic deletion of 17p11.2. She then followed up with a paper in 1986, in collaboration with Dr. Ellen Magenis, in which they described the clinical phenotype in 9 patients with the deletion. Since that time, Ann has devoted much of her professional career to furthering our understanding of this complex neurodevelopmental disorder.

In addition to those who have focused on particular diseases and syndromes, there are also genetic counselors with expertise that crosses the boundaries of professional practice to benefit us all. I think about Joy Larsen Haidle up in Minnesota. Joy’s a cancer counselor, and among her many talents, she has become an expert in medical billing and reimbursement for genetic counseling services. Joy is a past NSGC board member and has been involved in many

payer initiatives. Along the way, she has learned more than she ever wanted to know about billing, reimbursement, and coding. I have no doubt that when she's not sharing all this important knowledge with the rest of us, Joy is very successfully getting payers to cover her services up in Minnesota. Dan Riconda is another genetic counselor whose knowledge about state licensure has benefited our entire field. Dan is down in Florida which was one of the first states to try to pass a genetic counselor licensure bill. Dan spent countless hours on the road between Orlando and the state legislature in Tallahassee, grooming his skills in an area that he never thought he would. Dan has truly become a licensure guru, and he has worked closely with NSGC to disseminate this information to licensure committees in other states. Many genetic counselors have been mentored by Dan about the ins and outs of state licensure, and thanks in part to his perseverance and expertise, dozens of states have now either passed bills or are on the path to achieving licensure. The irony of all this is that Dan's home state of Florida still does not have licensure for genetic counselors. So here's to you, Dan Riconda, for sharing your wisdom. We're all hoping that someday soon, Florida will pass a licensure bill!

Elissa Levin is a genetic counselor whose pioneering work at Navigenics has changed the way we think about service delivery models for genetic testing and counseling. She is an industry innovator who continues to chart new roads as we move into the era of personalized medicine. Along the way, because of Navigenics' novel service delivery model, Elissa became a go-to person for the media when they wanted insights on the future of genetic testing. As a result, Elissa has made some very high profile television appearances, including segments on the *Dr. Phil* show and *The Doctors*. When Elissa is out there in the public eye, in front of millions of people, she describes herself as a genetic counselor, and she represents us so well. So here's to you, Elissa Levin, industry innovator and occasional TV personality. We may not all be TV stars, but your work in getting the word out to millions of people helps move our profession forward.

Julianne O'Daniel was here earlier this week speaking about next generation sequencing and its many implications for patients and professionals. She has been at the forefront of genomic medicine, trying to anticipate how this will all play out in terms of genetic counseling. When she is not

presenting, publishing, or thinking deep thoughts, Julianne is an incredibly organized and efficient NSGC volunteer. She is also a pioneer to watch as we move toward personalized medicine, an area of huge importance and promise for our profession.

I'd also like to highlight a whole group of genetic counselors at the Cleveland Clinic's Genomic Medicine Institute. I had a chance to visit them a few months ago, and I was overwhelmed by their energy, enthusiasm, and wide range of professional interests. I think of those genetic counselors as a reflection of our field and how far it has expanded into the larger genetics universe. The Genomic Medicine Institute recently built a new facility that includes a wing, which they affectionately call "genetic counselors row", where this large team of genetic counselors is housed. They're doing pediatrics and cancer counseling; they're involved in esoteric areas like genetic ophthalmology; they're publishing research and gearing up for genomic medicine; they're looking at health IT and how genetics will fit into the electronic medical record; they're active members of NSGC and hold leadership roles on our Special Interest Groups and committees. When I went to see them, I entered the building, and there was a palpable energy emanating from genetic counselors row. It was amazing! There were beams of light shooting out of the doorways. I had to wear special sunglasses to keep from burning my retinas. But perhaps the most remarkable thing about all this fabulousness is that it's happening in *Cleveland*.

Finally, I want to thank you again for selecting me as your 2012 NSGC leader. As I look out at all of you, representing a wealth of different specialties and practice areas, I am proud that NSGC, our professional society, has been there for us every step of the way. Our field continues to work hard to promote gender, racial, and cultural diversity, and we still have a long way to go in those areas. Perhaps because we're genetic counselors, we've overcompensated for these shortcomings by having an abundance of *professional* diversity. So the next time you find yourself thinking, "Have I strayed too far from the field?", "Am I still a genetic counselor?", remember your core skills. These are the fundamental attributes that identify us as genetic counselors, but it is our experiences, our creativity, and our passions that diversify us. Thank you very much.