Keynote

You can view this presentation at: youtu.be/ZJ7Nf1Rke-M

[00:00:05] **Dr. Sandy Siegel:** Before I start here, I wanted to thank the staff of the Siegel Rare Neuroimmune Association. They have not been recognized. And I'm going to start with -- it absolutely blows my mind that we have a staff. When we would do mailings, my parents would sit at the kitchen table with me, putting labels on envelopes, thousands of them, and we would fill up the floor in my garage. And there was a wonderful postmaster where we would mail out of in Powell, Ohio, who would come on a Sunday morning to help me load up the back of her truck to take these mail sacks to the post office.

[00:01:02] When I say, "We were a mom and pop operation," we were a mom and pop operation for a very long time, and for us to have the staff that we do, it's remarkable to me on so many different levels. But there's such a great group of people. I loved working with all of them, and if you're able to stand, please stand up. If you're not, raise your hands. I want to make sure the staff gets recognized. What an incredible job you guys have done. To put on a program like this just takes such an enormous amount of work, and I also want to thank all the medical centers who were involved in helping to put all of this together. Thank you. Thank you. Thank you.

[00:02:18] The other thing I want to say is, we have no control over what doctors decide they want to specialize in the rare neuroimmune disorders, so it's as likely that we would get a whole gaggle of jerks and jackasses as we would have some really exceptional people. And I have to say, we have been unbelievably blessed with the most amazing human beings across the board. So, thank you all for being involved in our community in the way that you are and offering the exceptional care that you do to the patients in our community. It's appreciated. So, thank you.

[00:03:18] Also, I want to remind everyone, we're in a room that doesn't really have a gazillion people in it. We have people from our community on Zoom, and as importantly, every single one of the presentations that you guys make over this weekend get uploaded to our website. It has been that way since 2002. All those videos are on our website from the very beginning when we started these education programs, and we do not provide medical advice because we don't have the training and experience to do so.

[00:04:01] So, somebody calls and says that they have issues with bowel, or bladder, or spasticity, or fatigue, we send them to the website to look at all of these videos that you guys provide. And they are seen thousands,

and thousands, and thousands of times. It's miraculous how many times those videos get watched. So, the impact that you have in providing this information to the community goes way beyond what happens in this room, and we are eternally grateful for it. Thank you.

[00:04:44] In 1994, as I said, we were a very mom and pop operation, and when Pauline diagnosed, there was nothing for the patient community. There wasn't the Internet in 1994. There were no specialists. There was absolutely no research going on at all. There was no support network. We really felt like we were out on an island, alone, and we were. The association got started -- that is another whole long story that I'm not going to get into -- but we had our first education program in Seattle in 1999. And the chief resident of neurology from Johns Hopkins came to that program, did a presentation there, and part of what he was doing is trying to figure out what he wanted to do when he grew up.

[00:05:58] After being with us for that weekend, which was an absolutely amazing, amazing weekend, Brian Weinshenker presenting his plasma exchange results; it was remarkable in so many ways. Doug Kerr, went back to Baltimore and announced that he was going to start a center of excellence at Hopkins with Carlos Pardo and with David Irani. And by doing so, he changed the landscape for people who have rare neuroimmune diseases all over the world. And he was doing it all. He hung up his shingle, and as a result, and through our promoting that Doug was doing this, he was getting patients coming from all over the world because there was nobody else who had the shingle hung up.

[00:07:03] And as a result, Carlos, David, and Doug, learned about transverse myelitis from that experience. In addition to the clinical care that he was providing to our community, he was also training physicians, and Ben Greenberg was one of his fellows, Michael Levy was one of his fellows, and there have been many more. And he was doing the first systematic research on transverse myelitis. He's an MD-PhD, and I didn't even know there was such a thing. He was doing basic research, he was doing research on restorative therapies, he was doing it all.

[00:08:01] And he was also getting other physicians interested in the topic by getting people together to develop the clinical criteria for diagnosis of transverse myelitis, which also made other research possible. And shortly after he started the center, he got a grant from the Transverse Myelitis Association to hire a research administrator who had just graduated with a master's from the Bloomberg School at Hopkins, Chitra Krishnan, who is now our Executive Director of the Siegel Rare Neuroimmune Association.

[00:08:58] Doug was putting on education programs, we were collaborating to do that, and he was bringing scientists together at meetings to review all of the research that was going on with the rare neuroimmune disorders. And he would come to all of our family camps, and he was the biggest kid at camp. And is important to me, he became a dear friend to me, and to Pauline, and to Pauline's service dog, Kazu. He had a very interesting relationship with Kazu. So, it is really my great honor to introduce our keynote speaker tonight, Dr. Douglas Kerr.

[00:10:02] **Dr. Doug Kerr:** I love you.

[00:10:03] **Dr. Sandy Siegel:** Love you too.

[00:10:04] **Dr. Doug Kerr:** Well, Good evening, everybody. Thank you for having me. I'm so excited to be here. The first transverse myelitis symposium that we had was in Baltimore, was in 2004, so 20 years ago. I've been involved in this community, as Sandy has said, since 1999. And it has been the greatest honor of my career to have been a part of this community, to advance this community, to learn and be a part of great people. And I want to tell you a few things about that journey along the way.

[00:10:38] I am an MD-PhD. I was trained in neurology with great colleagues like Carlos Pardo here and David Irani, and we went through residency together. That's where we learn about neurology. We didn't learn much about these diseases, I have to say. We learned very little about them. But we saw patients with transverse myelitis, or optic neuritis, or ADEM. And we were. I remember conversations with Carlos, being very, very, intrigued by what would trigger these rare neuroimmune disorders. And the hallmark of what an immune system should do is to attack foreign things and to ignore your own body. That's what the immune system does.

[00:11:37] It recognizes things that are not you. But in an autoimmune disease, and in a spectrum of autoimmune diseases, including these, the immune system gets tricked. And so, it reacts against your own body. And where that attack occurs is really what, in some ways, differentiates optic neuritis, ADEM, TM, etc. So, it was fascinating for us to think about how the immune system could go so wrong and to start to understand why that is. And we've begun that journey. And we've begun that journey starting with the Transverse Myelitis Association, now the SRNA, starting with the Johns Hopkins Transverse Myelitis Association.

[00:12:40] And now, where we've gotten to are people throughout the world, experts in the care of these disorders, and the research, and understanding of them, and developing new treatments. And it's been a phenomenal journey where people now understand the diagnostic criteria, which we set up initially in 2002. You couldn't even define what these disorders were. And we set up rigorous criteria. Hiring Chitra was one of the most important things this community did was because we knew that we didn't know anything. And we knew that we had to learn.

[00:13:30] So, we put up a shingle and said, “We are the Johns Hopkins Transverse Myelitis Center.” What we didn't tell you is, we don't know anything about it. And for the first 50 patients, we didn't, but we gathered information. And it's one of the long-lasting lessons I've ever had is, from then on, but initially, it was so important to just listen -- what were patients telling us. And it was an amazing experience. And we would leave our clinic days at Johns Hopkins, and we'd say, “My God, we learned more from them than we taught them or did anything for the patients.” But it forged this incredible alliance of the community, and that's a lesson that has been with me for my entire career.

[00:14:31] It is the patient, the caregiver, the advocate, the scientist, the clinician who all must bring their all to this to advance the field. And that's what's happened. And if you look at the SRNA now, and you look at the fellowships, which are designed to train tomorrow's physicians and scientists about these disorders -- we've been doing that for a long time -- and it's changed the landscape. You know about the connection aspect of SRNA, which is so important, to talk to other people, to be in this type of a meeting where you're learning from other people, other caregivers, you're learning from scientists, and just like in the beginning, you were teaching us at events like this.

[00:15:32] So, it has been an amazing journey. It's not just the fellowships. Sandy mentioned the camps, which, if you ever get a chance to go, are life changing. Unbelievable camps where patients, siblings, caregivers, mothers, fathers get together and just play, and swim and go up in hot air balloons, and we dance a lot. I don't dance well, but I dance a lot. Wait, did I hear something from over there, Ben? And so, it changes the lens through which you see individuals and through which they see you. And that's really, really important.

[00:16:31] Now, I won't walk you through all of the other things that SRNA does, and I'm still obviously very involved with SRNA, and we still continue to expand the potential of what this community can do. And I'll say one thing is that, we're just at the beginning. So, it's taken time. It's taken this time from 1994, 1999 is when I got involved, up until now, to really gather everything that we need to make a dramatic change. And I'm talking restoration of function, and I'm talking prevention, and I'm talking cures. And that's where we are poised to go.

[00:17:22] We couldn't without the SRNA, we couldn't with all of these clinicians and scientists, and we couldn't without learning from you. So, I want to tell you one more thing before I finish, and it's not related to rare neuroimmunological disorders. So, I've spent the last 15 years of my career doing something else. I moved to Cambridge, Boston, and I began to work for, and then create biotechnology companies to develop new therapies for patients. And that's what I've been doing since roughly about 2006. And I want to tell you about one experience on that.

[00:18:15] So, back when I was at Johns Hopkins, I studied a disease called spinal muscular atrophy. It's a baby disorder. It's a genetic disorder. Babies are born normally, but then, by six months of life, they get weak. They can't suck, they can't feed, and they die by 18 months of life. That is spinal muscular atrophy.
[00:18:44] My laboratory at Johns Hopkins was studying motor neuron disorders, the neurons that allow people to move and to have strength. And in this particular genetic disorder, those spinal cord cells die, and it's a genetic disease. And we and others, back in about 2002, made the recognition that there is potentially a backup gene that you could activate to impact the symptoms of babies with spinal muscular atrophy.

[00:19:20] It came out of a bunch of different laboratories, and we started to test it in animal models, spinal muscular atrophy, and we would deliver this therapy that would activate the backup. And guess what it did? It worked in the animals, and it made them stronger, and it protected their neurons. And so, now, I'm up in Cambridge, in Boston, and I'm at a company called Biogen. And I said, “We're going to do this at Biogen. We're going to develop a drug for babies with SMA.” And we did.

[00:20:03] And oh, did we. And we started giving it to babies, and what happened is, they actually didn’t progress anymore. They did not progress to being on a ventilator. They did not die. But they also didn’t really recover. And we thought we knew why, we do know why. By the time symptoms present, there has already been damage.

[00:20:35] So, when we’re taking individual’s babies who are already weak, there’s only so much you can do. So we asked ourselves, “What could we do to get to patients earlier?” And the answer was, “What if we could screen babies at birth? What if we could screen babies at birth for whether they would get SMA?”

[00:21:04] And we developed a test, and we did this with the CDC. And I traveled around the world to try to get people to pilot this newborn screening test. By the way, the test costs one penny. Very cheap, very easy. And when we piloted the test, if any child was diagnosed with what would become SMA, they were given this experimental therapy, or the option to take this experimental therapy.

[00:21:39] Now you can imagine that’s a tough conversation. Two parents who are normal, it’s autosomal recessive, they are carriers, they do not have the disease. Baby, normal at birth. Maternal SMN protein, baby doesn’t get symptoms until 6 months of life. But you’re telling these parents that their child is going to die by two years of life even though the child looks normal, and that we have an experimental therapy that we don’t know if it works, but we’d like to offer it to you. A tough conversation, right?

[00:22:21] But we had those conversations throughout the world. Taiwan, France, many states in the U.S. Most people did not want to do this newborn screening test because they said “What are you going to do? If you diagnose this baby, what are you going to do? You don’t have a therapy.” Well, we think we do, it’s experimental, we think we do wasn’t enough for some places, but it was for others.

[00:22:45] Okay, so let me cut to the chase. So, any baby genetically diagnosed with what would be SMA got offered this therapy, and now, we’re thirteen years later. None of those babies has any disease at all.

[00:23:12] So, full stop. Totally normal, running, playing, kicking balls. Thirteen years old. No disease, at all. As a result of that, this drug was approved. It's now called Spinraza. It's available throughout the world. As a result of that, newborn screening was augmented to include SMA and babies in every state in the U.S. are now screened at birth for SMA. And if they are genetically identified as going to get SMA, they are treated and the disease is going away. Because nobody gets it.

[00:24:04] So why do I tell you that? Because that's what we want to do with rare neuroimmunologic disorders. And it's time. And we couldn't have until we got to this point where we've got people in this room, and scientists, and clinicians, and patients and caregivers telling us. And the medical community listening.

[00:24:37] And you've got guys like Ben Greenberg doing trials with Q-therapeutics trying to repair the spinal cord here at UT Southwestern. You've got guys like Michael Levy really looking at the genetics, or "Why does the immune system go wrong, and why is it tricked?" And Michael is looking into that. So we're poised for this. Which is not to say that a diagnostic test or a preventative test is going to be easy, and it's not going to be tomorrow.

[00:25:15] But to be a part of this community that is moving in that direction is incredibly satisfying, and I'm glad to be a part. And I thank you for coming here.

[00:25:27] And I thank you for taking some time. (Applause)