# The Rare Disease Gazette

*Conversations with the world's experts about rare disease*

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# BUILDING STRENGTH TOGETHER

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# Editorial

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Rare Disease Detection: Rare But Not Alone

The plight of patients with rare diseases is a critical unmet need of patients in healthcare. The statistics are frightening; there are 7000 rare diseases in the world that affect 350,000,000 people. One in eleven Americans has a rare disease. Three-quarters of patients with rare diseases are children and only half of patients receive an accurate diagnosis. The average delay for a patient to receive a diagnosis with a rare disease is 1 1/2 years. It is deeply concerning that one in four patients with a rare disease waits four years for an accurate diagnosis. There is an urgent need to communicate knowledge and expertise in the field of rare disease detection.

The journal *Science*, (American Association for the Advancement of Science) in collaboration with Fondation Ipsen delivers international science webinars for the general public.

*The Rare Disease Gazette* is a magazine that broadcasts these discussions.

James Levine

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## DON'T MISS!

### The Conversation

**Experts of the month:** Erika Berg, Ph.D, hosts a conversation with world's experts about rare disease: Building strength together: How rare disease caregivers form communities to support each other and their loved ones.

# The Conversation

### Experts of the month

**Marissa Bisho, M.A.** *(CDKL5 in Color, Connecticut, USA)*
**Elle Cole, B.A.** *(The CleverlyChanging Podcast, Maryland, USA)*
**Jan Domaradzki, Ph.D.** *(Karol Marcinkowski University of Medical Sciences, Poznan, Poland)*
**Jennifer Siedman, M.Ed.** *(Courageous Parents Network, Massachusetts, USA)*

**Erika Gebel Berg, Ph.D.**
*(Science/AAAS, Washington, DC; moderator)*

***"You can listen to a podcast no matter what you are doing, as many of us caregivers are often multitasking. I saw it as a way for me to take control of something in this chaotic, rare-disease space. When you have a genetic disorder, there are different emotions and feelings that go along with that and I said to myself that I cannot change the past, but what I have control over is the future and what other parents and families know about this disease."***

**– Elle Cole**

**Erika Berg (host):**
*Today we will explore the diverse strategies caregivers use to form and maintain communities, both in person and online, and how these communities provide critical, emotional, informational, and practical support. We will discuss the similarities and differences between in-person and virtual networks, highlighting the unique benefits and challenges each offers. Additionally, the panel will address important mental health issues in the caregiver experience, examining how caregiving can affect mental well-being and how community involvement can provide the necessary support to prevent burnout and foster resilience. I would now like to take the opportunity to welcome a truly distinguished panel. I will give each of them a chance to say hello and introduce themselves.*

**Elle Cole:**
My name is Elle Cole. I am a caregiver and a mother to twins. My daughters are 16 years old. One of my daughters has a rare disease called sickle cell disease. Sickle cell disease is one of the most common genetic disorders. However, in the United States, fewer than 200,000 people have this disease, which is why it is considered rare. I am also the host of a podcast, and I am the marketing manager at a sickle cell organization. Thank you for having me.

Marissa Bishop:

My name is Marissa Bishop. My background is in social work. I was an elementary school social worker for many years before I had my son. My son Gregory lives with a rare disease called CDKL5 deficiency disorder. It is a developmental and epileptic encephalopathy, which means that he lives with multiple profound disabilities, medical complexities, and a very significant epilepsy disorder. I spent four years volunteering, leading fundraising and community engagement for a CDKL5 foundation, and now I produce and co-host a podcast for our community called *CDKL5 in Color*, where we explore the different symptoms of the disorder, and we welcome our peer caregivers to come on and share their lived experiences.

**Jennifer Siedman:**
My name is Jennifer Seidman. I am a mom to three. My middle son, Benjamin, was diagnosed in 1996 with a lysosomal storage disorder called Sanfilippo syndrome. It is a neurodegenerative disease. If you had met him when he was two, he would have seemed like a typical two-year-old, but by the time he passed away at the age of 17, he was no longer able to walk, talk, or swallow. My caregiver experience changed over time and became more intense. A year after Benjamin passed, I got involved with an organization called Courageous Parents Network, where I now serve as Director of Community Development. Courageous Parents Network is a nonprofit organization that provides digital resources and programming to help caregivers, and others caring for children with serious medical conditions, to navigate the illness journey with support and a sense of community. At the center of Courageous Parents Network is a video library of over 700 videos featuring the voices of parents and clinicians to illuminate that shared experience and to also help to reduce the isolation that many caregivers experience.

**Jan Domaradzki:**
My name is Jan Domaradzki. I am an assistant professor and medical sociologist at Poznań University of Medical Sciences in Poznań, Poland. I have been researching the social dimension of rare diseases for almost 15 years, with a particular focus on family caregivers. Over time, my research became more personal as for the past 12 years I have been a caregiver for my son who lives with an intellectual disability and autism. Thus, although our experience is fairly different than that of caregivers of individuals with rare diseases, many challenges and needs overlap. In my scientific work I try to advocate for caregivers' needs with my evidence-based engagement.

**Erika Berg (host):**
*I am going to put my first question to Elle. Can you share with us some of the unique day-to-day challenges that rare disease caregivers face?*

***"When you have a child that has a rare disease, life is unpredictable. You never know when things are going to change."***

**Elle Cole:**
One of the day-to-day challenges is living in fear of what can happen next. When you have a child that has a rare disease, life is unpredictable. You never know when things are going to change. It means be-

ing observant and watching your child. It is also being present in the moment, as you do not want the child's rare disease to consume every moment. You want to be patient. You want to be a loving parent, but you also want to be on top of the medical complexities that they encounter. It is a challenge, to be all of those things. Sometimes you have to be a mathematician or a medical professional. You wear so many different hats. That can be unexpected, as many of us did not go to medical school. Suddenly, we are playing catch-up, doing research, and becoming experts for our children.

**Marissa Bishop:**
I feel exactly the same way. I think that unpredictability is really unique to the rare disease caregiver experience. You have to learn to accept that as your reality. You could have all these beautiful plans for your day or night, and imagine how you think it will go, but the reality is that you are not in control of how it is going to go. Medical things can change on a dime. My son has seizures every day and they can occur at any time. Depending on their severity, they impact what is happening for our family. So, I think that unpredictability and learning how to navigate it in your life is really unique to a rare disease caregiver experience. It is really difficult. It is part of the reason why I am no longer able to work. I have to be available for my son no matter what.

**Jennifer Siedman:**
We hear this all the time at Courageous Parents Network from the families that come to us. They live in what I like to call a chronic crisis existence, where you do not know what is going to happen next. Maintaining that level of hypervigilance is exhausting. I think we forget to think about the exhaustion factor that comes from so much of what we have to do. The world is better at recognizing the physical or visually apparent factors, but less adept at recognizing the mental exhaustion that comes on when we live through these experiences.

**Erika Berg (host):**
*Jan, over the past decade, you have done a lot of research into the needs of caregivers for people living with rare diseases. Based on what you just heard, could you share some of your findings in terms of what issues caregivers face and what broader impact that can have on their well-being and mental health?*

> ***"Our research confirmed that although rare diseases differ significantly in their epidemiology, etiology, symptoms, and prognosis, most stressors or challenges faced by caregivers are quite similar and include physical strain, emotional exhaustion, and feelings of loneliness."***

**Jan Domaradzki:**
In recent years, I have conducted several research projects on specific rare including Dravet syndrome, Angelman syndrome, 22q11 deletion syndrome, and Duchenne muscular dystrophy. We have also conducted two large nationwide surveys, each of which included over 950 caregivers providing care for over 1000 children with rare, ultra-rare, and even hyper-rare diseases. Our research confirmed that although rare diseases differ significantly in their epidemiology, etiology, symptoms, and prognosis, most stressors or challenges faced by caregivers are quite similar and include physical strain, emotional exhaustion, and feelings of loneliness. What is important is that family dynamics, both in terms of the relationship with a partner or spouse, and with the healthy children, are seriously affected. Caregivers frequently do not have time for themselves. There is a serious gender gap. We find it here in Poland, but it is also typical for many other countries. For many reasons, including cultural ones, it is frequently women who devote themselves to caregiving, which can be 24/7. They often have to quit their job entirely, which is of course also quite challenging for them. One of the most important challenges is their experience or contact with the healthcare system, which is frequently very overwhelming. Many caregivers feel that they are invisible to the system because in many countries, including Poland, healthcare professionals frequently focus on the disease or on the patient themselves, while the caregivers' needs are overlooked. To give you a couple of numbers, for example, our research shows that up to 90% of caregivers experience chronic stress, 77% experience depression, and 66% experience anticipatory grief, which Jennifer also mentioned and is a form of chronic stress. Many feel lonely and misunderstood by those around them, even by their closest family and friends. So, what is important to remember is that caregiving for a person with a rare disease creates a multidimensional burden that affects the physical, mental, psychological, emotional, and social health of caregivers.

**Erika Berg (host):**
*Marissa, you recently wrote an article for Rare Revolution Magazine describing your experiences in trying to find community as a parent and caregiver for a child with a rare disorder. Can you talk about those early days and some of the challenges you faced in getting the support or connection you needed from families that were not grappling with those issues?*

> ***"I found it really difficult to connect to the moms who were having a more traditional parenting experience. So, it was very isolating, not only socially, feeling like I was not able to connect with them, but even physically."***

**Marissa Bishop:**
When my son was born, I did not know that he had a rare disease. In the grand scheme of things, we were lucky. We got a relatively early diagnosis. He was diagnosed at six months old and I know that many people have to wait many more years. Initially, I did not know what was going on with him. He started having seizures at four weeks old, but like a lot of new moms, I had wanted to make new mom friends and had joined some of the Mom and Me groups in my local community. It became evident really quickly that it was going to be harder to build those relationships than I had anticipated. I would go to the groups and as the other moms wanted to talk about milestones or sleep training or things like that, I was wondering if I needed to switch neurologists for my baby and if I should be starting feeding therapy and early intervention. I found it really difficult to connect to the moms who were having a more traditional parenting experience. It was very isolating, not only socially, feeling like I was not able to connect with them, but even physically. I would be in the same space as these

women and as their babies started to grow, develop, crawl, roll, run and move, the moms were busy with their babies. At the same time I was still sitting in my same place, cradling my toddler and giving him a bottle because he was not able to eat food orally. As time went on, the disconnect between our experiences grew wider, and I found myself no longer willing to try to fit in. That is when I had to pivot and find other mothers who were living a similar experience as I was.

**Erika Berg (host):**

*Jennifer, when you first assumed the role of caregiver in the rare disease community, how did you figure out where to turn for advice, support, and guidance?*

**Jennifer Siedman:**

When Benjamin was diagnosed in 1996, the internet was fledgling. There were no social media groups that you could join and finding families that were on a similar path was very difficult. Like Marissa, I also felt separated from the typical mom. I could not stand in the pickup line at elementary school and talk about my experiences with Benjamin with the other parents, because they were so disconnected from the typical parenting experience. You had to make an effort to lean in and go to patient organization conferences. Those could be scary too because sometimes while you may feel embraced by being around others that know and see you, you also see into your future or what might be coming. That can also be emotionally challenging. One of the things I love about Courageous Parents Network is that you can go on the website and watch a video of another parent talking about their experience. If it is not resonating with you, you can simply turn it off and pick a different story to listen to, unlike at a live conference. Live conferences can be great, but sometimes you have to be in that same space with everyone. So, I like to think of Courageous Parents Network as a place where you can "try on" the caregiver hat that works best for you by listening to different stories and hearing about the similar experiences and emotions that you are encountering along your path. Unfortunately, Courageous Parents Network did not exist when I was caring for my son, Benjamin. It was only a year old when I got involved. So, I have helped build the organization that I see is now impacting not only families of children and caregivers, but also the clinicians who support us along this journey by caring for our children and hopefully caring for us as caregivers as well.

***"For my very first Mother's Day, my husband brought me a computer and said, "I think you should blog." I did not really know much about it, but I love to write. So, I said, "Okay, the community does not exist, so I am going to create it." I started writing my story and sharing my experiences. I then started to find other parents who would sometimes comment, and they would connect with me. Even though our stories were somewhat different, there was a commonality there."***

**Elle Cole:**

What you said really resonated with me because when my daughter was diagnosed, it was a unique experience, as there was no one else in my family who had sickle cell disease. I could not explain it well enough and even though I tried, they did not necessarily understand. I was not an online media person and I did not have social media at the time, but that was really the door I opened to start embracing the idea of finding a community. I actually started with my blog, cleverly-changing.com. For my very first Mother's Day, my husband brought me a computer and said, "I think you should blog." I did not really know much about it, but I love to write. I said, "Okay, the community does not exist, so I am going to create it." I started writing my story and sharing my experiences. I then started to find other parents who would sometimes comment and they would connect with me. Even though our stories were somewhat different, there was a commonality there. I was willing to be open in order to get to know other people. My daughter was diagnosed at newborn screening, so I knew almost immediately as they confirm it at the three-month mark. I received a letter in the mail and the letter stated, "Your child has hemoglobin SS." It was such a cold and isolating experience. Even though I have a husband who is very supportive, I felt like I was experiencing everything alone. I knew that if I stayed in that state of mind, I would not be productive. I remember the day I got the letter, I looked down at my twin girls as I was feeding them and I remember seeing my daughter smile. That was really the light bulb moment that said, "You can do this. You can figure this out. You can try." And what trying looks like is different for everyone, but that is when I had the courage to not give up and to get out of the house. I started to open up and be honest with myself that this was a real diagnosis. Sometimes I think at the beginning, you are in denial. Specifically with the rare disease my daughter has. Until she experienced her first pain crisis, I think I was in denial for some time. During her first crisis, when the blood starts to sickle and it becomes hard and sticky, she was in a lot of pain. It was not something that I could change. I could not control it. I could not remove it. I gave her the medication like the doctor said, but it was still there. That was when it became real for me and I found other parents by talking to the doctor. They said these support groups exist, but I never could find those particular support groups. I went online and found people who were like-minded and, like Jennifer said, I started to go to conferences. My first conference was in 2019, when my daughter was 11. It was an eye-opening experience, and I felt like these were my people. I felt like it was a family reunion and I felt accepted. That was when I decided that advocacy was something that I would continue and become a part of, and I would usher in new parents. I would get to know them so they did not have the same isolating experience as I had. That was really a groundbreaking moment in my family's life.

**Erika Berg (host):**

*Jan, from a research perspective, have you looked at what the newly diagnosed entering the rare caregiver community are typically doing in that space when they are trying to reach out? Or is that an unknown?*

***"Although in some cases, physicians serve as types of guides who point to a particular patient association or foundation, in most cases, families found themselves navigating their reality totally alone."***

**Jan Domaradzki:**
What was just said is also reflected in our research, as well as in global studies. Due to the rarity of these conditions, most caregivers describe the initial post-diagnosis phase, like Marissa did, as marked by confusion, fear, loneliness, and lack of direction. Although in some cases, physicians serve as types of guides who point to a particular patient association or foundation, in most cases, families found themselves navigating their reality totally alone. For example, nine of the 11 associations I am collaborating with said that they had to create their own association because there was nothing that existed. The key turning point frequently comes through online networks, such as online support groups or social media pages, or sometimes through a chance encounter with another parent, for example on the hospital ward. This often leads to creating or joining patient advocacy groups because caregivers frequently describe their experience as a lonely ride, where they are left by themselves and must unite without the official support from the state or other agencies.

**Erika Berg (host):**
*Marissa, in that same article I mentioned before you talked about how you had mixed feelings around traditional social media networks like Facebook. Can you share how you feel about those social media networks, including the pros and cons, and let us know where you ended up finding your community?*

***"I feel like Facebook is the go-to place for community support for rare diseases, especially the extremely rare diseases. It is free, it is accessible to almost anyone, and if you have good moderators, it can really be a positive experience to be a part of those groups."***

**Marissa Bishop:**
When my son received his diagnosis, the genetic testing was ordered by his neurologist. When it came in, the neurologist called me on the telephone. I remember he said, "We know why your son is having seizures. He has a mutation on the CDKL5 gene." And I am trying to write it down: CDKL5. I had never heard of anything like that before and he said, "We can talk about it more, but I do not know a lot about it." That was the end of the phone call. The first thing I did is I went on Google and I typed in CDKL5, and there was a foundation that already existed. I do not know if my neurologist had checked Google before he called me but I wish he would have told me, "There is a place you can go to find other people." So, I found our foundation through Google.

The next thing I found was the Facebook support group. Our Facebook support group has been in existence for over 10 years and there are pros and cons to social media. I feel like Facebook is the go-to place for community support for rare diseases, especially the extremely rare diseases. It is free, it is accessible to almost anyone, and if you have good moderators, it can really be a positive experience to be a part of those groups. You are sharing photos and stories, you are asking questions, and you are receiving immediate feedback. I can post a question in our Facebook support group and hear back in an hour from multiple people, as opposed to trying to ask my doctor and wait days to hear back from him. Of course, I would not take medical advice from the people in the Facebook group, but if you have something on your mind, you can get feedback pretty quickly. I think other social media, like Instagram and TikTok, are fabulous for following content creators who reflect your lived experience. When I step outside of my house, I do not see other families that look like my family, but I follow a bunch of content creators on Instagram who are sharing videos of their kids and their day-to-day lives. When you can see somebody else having a similar experience, it really goes a long way in helping caregivers feel less alone. One of my favorite things about social media is the tips, tricks and hacks that you learn from other parents. This is the stuff that healthcare providers are not able to share with you. They just do not know the accessibility hacks, equipment hacks, or gift ideas, because buying gifts for kids with severe neurodevelopmental disorders is really hard. Being able to share, "I found this and my son seems interested," is really helpful. The biggest con for me is the fact that, especially with the Facebook support groups, those groups do not belong to the community. They belong to Facebook. Facebook can cancel them and delete them at any time, which would be devastating to communities, not only for the loss of the connections that you have made with other people, whom in reality you do not have another way to contact, but for the information that exists in those groups. For example, when my son's neurologist recommends a new anti-seizure medicine for him, one of the first things I do is type the name of the medicine in the Facebook search function in our group. I read all the questions and comments and experiences that people have shared about that medication. I am then able to take that back to the neurologist and say, "People are saying this about the medication. What do you think about that? Have you heard that? Have you experienced that?" There is over a decade's worth of that type of information in that group and it is an extremely valuable resource for families that does not exist anywhere else. However, it does not belong to the community and the families, and it is at the mercy of Meta.

***"In my community, there are so many different nuances that needed to be changed or updated and social media helped us to become more unified."***

**Elle Cole:**
I think social media really helped me develop a larger community where I started to meet people in my own state. Prior to social media, I just felt like everybody was so far apart. Once I started to attend those conferences, I started to meet people that were in my area and I started to be able to connect with them at home. Even though I do not necessarily see them, I am able to text them, like in a more intimate setting. I wanted to be able to learn how to share my story and to engage in legislative advocacy, and social media put me in touch with people who were already doing it. It made me feel that I could be a part of a larger system change. In my community, there are so many different nuances that needed to be changed or updated and social media helped us to become more unified. I also now work for a nonprofit in California that is related to sickle cell disease, and I would not have that job without social media because that is how they found me. We were on a board together and we were able to connect with each other. It not only widened my voice, but it helped me see that even though my area was not doing as many things as some of the others of the organization, we could create those things locally. So, it gave me

a vision of things to work towards and things that I could work with other people to create in my local community. It really just expanded our horizon and connected us all with each other. June 19th is World Sickle Cell Day and we have a 24-hour global marathon that I now get to help plan. I meet people from all over the world who have this rare disease and it has given me so much hope and courage about the future that I did not have before. For me, it has been one of those experiences where you start out with dreams and goals, but when you first get that diagnosis, it feels like they slip away. Then, when you have a community, all of a sudden it feels like you are being given that gift back. You are able to regain those dreams, regain those goals, or develop new ones that are in line with your future. So, I know that it is quite complex, but it has really been that way for me and my family.

**Erika Berg (host):**

*Jennifer, I want to talk more about the Courageous Parents Network as before you joined, you were involved with the network as a parent. Can you talk about how you benefited from connecting with Courageous Parents Network during a difficult time and what inspired you to eventually do the same for other families and continue on with this work as Director of Community Engagement? What is special about this organization?*

**Jennifer Siedman:**

First, I want to touch on something that both Marissa and Elle mentioned, which is the gift that the internet has given us and acknowledge that sometimes that gift can be a burden. One of the things that I also want to highlight, which is exceptional about Courageous Parents Network, is that clinicians come to us to understand the lived experience. I think the capacity of the internet now, through places like Facebook, TikTok and Instagram, allows us to show the world how we are living robustly and happily. Although there are a lot of challenges, we are actually living a life that is whole and good. I think it is important and it has provided an opportunity for those in the medical field to learn about our lived experience without having to be in front of that patient 24/7. So, you can now get your doctor to understand what your child looks like at home because you have the capacity to send a video.

As I said, when I first came to Courageous Parents Network, it was only a year old, and I did benefit from speaking about my experience of caring for my son Benjamin. My interview is with my husband and our palliative care doctor. We talked a lot about how palliative care changed the care experience of my son. He did not get a referral until he was 14 years old, and we were in a medical crisis at the time, which is another discussion, because I believe we should have been referred much earlier. I think our lives and our journey would have been much better if we had received palliative care sooner. It is one of the things we advocate for at Courageous Parents Network. I gained a whole new community after I started getting involved with Courageous Parents Network because, obviously, I have the opportunity to meet so many parents with so many different diseases. I get to see and study the similarities that we all share and have in common, because even though it is nice to spend time in your own disease group and with people whose diagnosis is similar, we all have so many shared experiences across the board. We have all experienced anticipatory grief. We all worry about maintaining our partnerships, whatever that looks like. We all think about how to have good doctor-patient interactions. These are all the kinds of topics we talk about on Courageous Parents Network. So, it has been nice for me to extend my relationship with my son Ben and continue my bond with him, so to speak, even after his death, through my work at Courageous Parents Network.

**Erika Berg (host):**

*Jan, do you know to what extent clinicians are accessing these online communities for information that they can bring into their practices? Is it a part of formal training or clinical recommendations, or is it more used on a case-by-case basis, based on what you have seen in your work?*

***"Research from many parts of the world shows that numerous healthcare professionals, including nurses, physical therapists, and physicians of various specialties, often lack knowledge and awareness of rare diseases."***

**Jan Domaradzki:**

Research from many parts of the world shows that numerous healthcare professionals, including nurses, physical therapists, and physicians of various specialties, often lack knowledge and awareness of rare diseases. Research also shows that even if they are aware of rare diseases, they frequently feel unprepared to provide care for such individuals and their families. It is for this reason that most associations, foundations, or patient advocacy groups try to educate not only parents, but also healthcare professionals. Most are eager to learn from the parents or the so-called "lay experts," because as we have over 10,000 rare diseases, it is impossible for physicians, whether they are general physicians or specialists, to be familiar with such a large group of diseases. They frequently acknowledge the expertise of parents, although, sometimes they neglect this knowledge. Most associations and foundations try to educate healthcare professionals, and many healthcare professionals try to use this expert knowledge to support their future patients. For example, they send patients or newly diagnosed families to different associations or organizations, and also try to participate in different seminars, webinars, and conferences. Each association or organization tries to have their own researchers who support their knowledge. So, there is a close established link between associations, parents, and the scientific community.

***"We spend a significant amount of time presenting at hospital organizations because we understand that if we can change the dynamic within the medical system, we can change the caregiver experience. It is a critical piece, and we are really setting out to transform care, both how it is given and received, by educating both clinicians and parents."***

**Jennifer Siedman:**

I would like to add that at Courageous Parents Network, while we are by parents and for parents as an organization, when we host our monthly webinars on

various topics, generally at least 50% of the people who attend are clinicians. They are coming to us to learn about the lived family experience through the programming that we provide and through the videos that we have. We spend a significant amount of time presenting at hospital organizations because we understand that if we can change the dynamic within the medical system, we can change the caregiver experience. It is a critical piece, and we are really setting out to transform care, both how it is given and received, by educating both clinicians and parents.

**Elle Cole:**
I also wanted to add that my daughter was born in 2008, and now I see an explosion of patient-centered organizations and community advocacy boards. You are now seeing a lot of hospitals that are asking patients and caregivers how they feel, and they are able to sit on boards and have direct input about the future and what needs to change. At first it was very different, but I am seeing more openness and willingness to do this. Often, the people who serve on these boards are recruited from other organizations in the community.

**Erika Berg (host):**
*Elle, you have written about the importance of mentorship for rare disease caregivers. Can you talk about making connections with experienced people who can help navigate the practical and emotional challenges of day-to-day life, and share some resources that could help others to establish those types of mentorship-like connections?*

> ***"Conferences are absolutely amazing, and I have met lifelong friends at conferences. I would say that if you are able to attend a conference, it is one of the best places to meet someone you can mentor and be mentored by."***

**Elle Cole:**
Let me start by sharing my story with mentorship. I mentioned that my daughter was diagnosed at birth with her rare disease, but when she turned six years old, she was also diagnosed with an autoimmune disease, called type 1 diabetes. When we went to the hospital for type 1 diabetes and we were in the emergency room, they gave me a paper and told me to sign up because I would be connected with a parent mentor. What was amazing about that is when she was released and I got home, another parent of a child with type 1 diabetes called me and said, "Right now it seems hard, it seems impossible, but you can get through this. You can learn how to give your child the medication that they need to live and do not give up." That was so unique because, again, this was not my first experience with chronic illness, but it was the first time I was immediately put into direct connection with someone who said, "You can handle this." And I said, "Wow, this is amazing!" I was also given what was called a bag of hope. In that bag of hope there was a teddy bear, books, and different things related to treatment, and I thought that was amazing, but it was not something that I received on the rare disease side. It was at that moment that I thought how wonderful it would be if this existed for everybody, so that you do not feel like you are isolated or alone in this experience. For me, it was an eye-opening moment where I began to write my book. I had already started teaching my kids, but I realized I needed to publish the books and the information I was sharing with them. I needed to create something for my rare disease community like the type 1 diabetes community gave me. So, I started to talk to the organizations and say, "This was my experience in this setting, what can we do to make it better?" We have tried to create what is called the Sickle Cell Caregiver Summit to give parents an opportunity to connect with other people. We do not yet have a system where you can sign up immediately when you are in the hospital, and I would love to get to that point, but we are not there right now. What I do, however, is actively work to create a space where parents can directly mentor each other. And that was because of my unique experience. To be completely honest, because my child had a dual diagnosis, I felt like I could not 100% relate to the rare disease community in the same way. I thought that she is going through all of these other experiences that are so different and unique, but now I realize that it is okay that we have some things that are different. We are all working towards keeping our children as healthy as possible and giving them the best quality of life, and that is what truly matters. When it comes to mentorship, it is something where you may have to reach out to organizations. I know it is hard when you are handling so much and trying to create your own organization, but sometimes that can help serve as the foundation of a mentorship. There are organizations like the Caregiver Network and the one I work for, which is called the Cayenne Wellness Center, where they will connect you with a support group. There, you can talk to caregivers or patients and just have a safe space to be honest and real. Those are the opportunities that do exist in the community. Global Genes is another community where you can connect with other people, and I know Jennifer mentioned conferences as well. Conferences are absolutely amazing and I have met lifelong friends at conferences. I would say that if you are able to attend a conference, it is one of the best places to meet someone you can mentor and be mentored by. It can be a cyclical relationship. It does not have to be one-sided. There are people who are giants in this community who have done it before, and you can lean on each other's shoulders.

**Marissa Bishop:**
I think the idea of having a mentor and having a mentor relationship is such a gift. It is a gift for the person who is experiencing the information and the support because this is new for them. As I have grown, and my son is 9 now, I have leaned on the mentors and the people in the CDKL5 community who have come before me. I am now also able to turn and share my knowledge and my support with other new families. So, you get to grow in that role from a mentee to a mentor, and it is really special. I think some of the mentor relationships that have been particularly helpful to me, outside of my son's rare disease, are with the local groups that help you understand how to navigate your local school system, your state waiver programs, and your state support services. It makes me feel a little angry that every single rare disease child in the country is seen by doctors and pediatricians, but we are not receiving that information about how to apply for our state waiver programs or how to apply for our state support services or the benefit systems for our kids. We are not getting that information from providers, and so you start to hear about it from other families. Then you start to say, "Well, what is this?" Sometimes you can be delayed in signing up for wait lists that you should be on, and it delays the support and services your family can receive. Those mentors who have been through your systems locally before, can turn around and say, "Hey, you need to sign up for this. You need to do this. Here is how I have navigated this before." It is incredibly important.

**Erika Berg (host):**
*It sounds like it is mostly informal, or have you had any type of formal pairing?*

**Marissa Bishop:**
I have not had any formal pairing. There are networks such as Parent to Parent USA. That is a national network that can help you connect to other parents. For me, it has been more informal. I have found that my child's therapists, both physical and occupational therapists, who also serve other families in my community, have been a really good connector. They say, "Oh, I know a mom. Let me ask if I can give you her number." They have been a really great resource to meet people.

***"As human beings we need anticipatory guidance. We crave to know what to expect when something like this is happening. We all probably went out and bought that book,* What to Expect When I'm Expecting*, when we had our first child. When your child is diagnosed with a rare disease, that book does not exist for you. You have to create it and put it all together by yourself."***

**Jennifer Siedman:**
I think what Marissa is bringing up highlights a big gap in our caregiver experience. As human beings we need anticipatory guidance. We crave to know what to expect when something like this is happening. We all probably went out and bought that book, *What to Expect When I'm Expecting*, when we had our first child. When your child is diagnosed with a rare disease, that book does not exist for you. You have to create it and put it all together by yourself. Go into the bookstore and look at how many books there are about typical parenting, and you will find a plethora of them. We are now hearing more and more stories from rare parents. We can go to places like Courageous Parents Network and listen to stories. We can go to conferences. We can lean on social media and mentorship to help us learn. However, all of that is really about seeking that anticipatory guidance, both medically for our children, and psychosocially and emotionally for the things that we are experiencing. I think one of the things we really need to focus on as a society is teaching people where to go to find it and even giving them the language to know what it is called. I did not know the word anticipatory guidance until I started working with physicians and the people at Courageous Parents Network. Now I think to myself, if I could have had all of that in the beginning, my care experience would have been completely different. If only I had known what to ask for. If we can equip people to be familiar the terminology and ask for the things they need, we help them build that caregiver muscle. I think Marissa brought up a good point about how we have to look in 90 different places to find that guidance. We have to go to schools. We have to go to groups. We have to go on social media. We have to go to a million specialists. It is exhausting.

**Erika Berg (host):**
*Jan, are there models for information sharing, whether it be through a mentorship or just a common source of information that brings everything together? And if so, what impact does having something like that have on caregivers?*

***"I believe that anticipatory guidance is not only needed by individual caregivers, but also by the different associations and foundations being launched by caregivers."***

**Jan Domaradzki:**
I would first like to add one comment to what Jennifer was saying. I believe that anticipatory guidance is not only needed by individual caregivers, but also by the different associations and foundations being launched by caregivers. Frequently, the associations that I collaborate with tell me that they have to invent the wheel over and over again. Not only do the parents have to search for all that practical information, but new associations launched by families and parents face the same challenge. It is also a particularly difficult task for them, because while providing care for their children, they must also gather all the information about how associations and foundations work. This is why anticipatory guidance is also important for associations, especially new ones that are being launched by caregivers.

Now going back to your question, there are, of course, several more broad or umbrella organizations. Global Genes, which was already mentioned, is an American organization, but there are many others such as EURORDIS, in Europe. Many of these organizations provide information on how to connect and how to access both medical and practical information. While the focus is often on disease-related information, caregivers frequently need very practical information regarding everyday activities, such as choosing the best type of wheelchair, and physicians are frequently not aware of such information. Guidance and mentorship is very important, although unfortunately most research shows that it is mostly informal. Very few places offer organized mentorship, like Parent to Parent USA. I think this is one of the most important needs for caregivers, parents, and association alike.

**Erika Berg (host):**
*In all of these spaces, people are sharing sensitive personal information. Marissa, what are some of the key factors that go into creating a safe and supportive environment in these settings, whether in person or virtually, so that caregivers feel comfortable opening up and discussing these personal details of their lives?*

***"In our group, there is this culture of everybody celebrating everybody. Our moderators do a good job to make sure that if there is a comment that comes across as particularly offensive or harmful, it is addressed right away."***

**Marissa Bishop:**
When I worked in the school system as an elementary school social worker, I would have small groups. We would always start, before we did any work together, by setting expectations and setting norms

for the group and understanding how we were going to speak and respond to each other, and what is appropriate and what is not. I think the same thing is important for any spaces where you are gathering families together. You want to provide expectations for how people will be addressed and respected and lay some sort of foundational rules for those interactions. I think that goes a long way in reminding people of the expectations. We also need to reinforce the fact that this is a nonjudgmental place when we are getting together. There is no playbook for how to manage a rare disease. People and families make decisions based on their knowledge, their culture, their experiences, and their situation. As long as no one is being harmed, there is nothing wrong with that. Everybody is trying to take care of their family the best that they can. Coming from a place where everybody understands the expectations and there is no judgment, I think that goes a long way toward helping people feel comfortable. We have this ongoing Facebook support group and I am in there every day and we also have moderators. I have heard horror stories from some other rare disease groups where they are not supported and the moderators do not really step in. In our group, there is this culture of everybody celebrating everybody. Our moderators do a good job to make sure that if there is a comment that comes across as particularly offensive or harmful, it is addressed right away.

**Jennifer Siedman:**
I think Marissa brings up a really good point. Being with others who understand your circumstance can be empowering, but the truth of the matter is that sometimes there is judgment and there is a lack of respect for an individual family's decision or the direction of care they choose. That is really unfortunate, but the reality is that in these social groups it happens. I think one thing that makes Courageous Parents Network so unique, and can make us seem a little more remote than a Facebook group, is that we are both decision- and disease-agnostic. You can come and explore the concepts that you are experiencing, such as how to care for your healthy children or anticipatory grief. We talk about interventions as well, such as how to decide if your child should have a tracheostomy or not. You can listen to different parents or clinicians talking about a topic, and find the one that resonates and helps you make the best decision for your child in an agnostic kind of way. Sometimes it is nice to go somewhere where you see people like you but not exactly like you, if that makes sense.

**Erika Berg (host):**
*Elle, you have a podcast. How did you come to recognize a podcast as a good vehicle for sharing stories and forging connections between rare disease caregivers, and can you share what went into the journey of building your podcast into a successful resource for the rare disease community?*

> ***"You can listen to a podcast no matter what you are doing, as many of us caregivers are often multitasking. I saw it as a way for me to take control of something in this chaotic, rare-disease space. When you have a genetic disorder, there are different emotions and feelings that go along with that and I said to myself that I cannot change the past, but what I have control over is the future and what other parents and families know about this disease."***

**Elle Cole:**
I started podcasting on my own podcast, called the *Cleverly Changing* podcast, as an outlet. Initially, I did not feel like I could send my daughters to school with other kids, and so I started homeschooling them, and I felt like it was a little isolating for me. I love people and interactions, it was a way for me to develop a larger community. It was a podcast where my kids could participate and were able to meet other kids. That is how that particular podcast started, but then I wanted to do a podcast that was more related to my daughter's rare disease, so I partnered with the Sickle Cell Community Consortium. They were very open to help me develop a podcast, which is the *Vitamin SC3* podcast, where we were able to talk to all different types of people about their different experiences. That really resonated with me because I grew up listening to radio. Talk radio was a huge part of my life, and I felt like it was a way to connect with people without judgment, because it is just listening to words. I knew that I had felt isolated at times during my experience, but a podcast can go beyond the four walls of a person's home. We are now a technology-centered society, you can listen to podcasts on your phone and you do not need to carry a boombox or a Walkman. I am really dating myself by saying that! You can listen to a podcast no matter what you are doing, as many of us caregivers are often multitasking. I saw it as a way for me to take control of something in this chaotic, rare-disease space. When you have a genetic disorder, there are different emotions and feelings that go along with that and I said to myself that I cannot change the past, but what I have control over is the future and what other parents and families know about this disease. The *Vitamin SC3* Podcast is broken into several segments. There is a caregiver segment, which I host, and then there is a science portion, a mental health portion, and a social portion, because we, as people, are doing so many different things. We have to celebrate all of it, just like Marissa said. I was able to do that with a trusted group of friends, and it has resonated with my community.

Now there are many different podcasts in my rare disease community, and people embrace them and learn. There is now a new rare disease podcast that I love about gene therapy, hosted by friends of mine. On December 8, 2023, the FDA approved two gene therapies for the sickle cell community, and that was revolutionary, but it came with a lot of trepidation and fear. That particular podcast shares stories from people who are interested in the therapy and the clinical professionals who are involved in making it happen. It really educates the community and ensures that the questions people have are answered. I love that I have been able to see people who have gone through the therapy, both those who have been successful and those who were not. Because there is so much new science and research, but it is not always successful and there are risks. The podcasts are honest about all of it, not just the positive parts. That is what I love about podcasts: you can be open and honest, and that is the beauty of them.

**Marissa Bishop:**
I have been doing community engagement for CDKL5 for a long time now, and I really recognize the importance of having multiple avenues that people can access content. Everyone has different learning styles and different ways they like to consume information. Starting a podcast for our rare disease seemed like a really fresh way to get the word out. Like Elle said, you just hit play on your phone

and whether you are doing dishes, folding laundry, drawing up meds, or driving in the car, you can listen anywhere. Other types of content, such as those on social media, are sometimes at the mercy of the algorithm, which determines if they are seen at all. Newsletters can also get lost in people's inboxes that get out of control sometimes. Rare caregivers are busy and life is unpredictable. With a podcast, you can hit play at the hospital, and you do not need to bring anything with you. It is just so easily accessible. What makes it even more meaningful for our community is the feedback we get. I receive messages all the time from caregivers saying, "That really resonated with me. I feel really seen." What is really special is when we have peer caregivers come on as guests. I have gotten feedback from them that their friends and family have been able to better understand them and what they are going through, in a way that they were not able to before. That is something that has been really special.

**Book # 12.29**

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