The Rare Disease Gazette

Conversations with the world's experts about rare disease

Issue #27 | Mai 2025

REDEFINING CONNECTION

FONDATION IPSEN PRESS

Editorial

by **James A. Levine**
MD, PhD, Président, Fondation Ipsen
www.fondation-ipsen.org

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

FONDATION IPSEN

DON'T MISS!

The Conversation

Experts of the month: Erika Berg, Ph.D, hosts a conversation with world's experts about rare disease: Redefining Connection: How young people are shaping the future of the rare disease community through technology and innovation.

The Conversation

Experts of the month

Richard Horgan, M.B.A.
(Cure Rare Disease, Woodbridge, CT)
Yamina Hsaini
(Yamina's Life, Paris, France)
Pablo Ramirez Uribe, M.Ed.
(Rare disease advocate, Bethesda, MD)
Shandra Trantham, Ph.D.
(Rare disease advocate, Gainesville, FL)

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

"We can make digital spaces safer for young people by creating truly inclusive content. For example, we should focus on normalizing life with rare conditions and help others understand what it is like to live with them. By doing this, we create spaces where people are more likely to support one another. Because the more people understand, the less they judge."

– Yamina Hsaini

Erika Berg (*host*):
Today we are exploring how young people are building vibrant and supportive rare disease communities in ways that previous generations may not have immediately recognized. Utilizing technology, social media, and other modern approaches, they are creating spaces for shared experiences, resources, scientific advancement, and advocacy that transcend geographical and generational barriers. These new forms of community are not only providing emotional support, but are also driving research, awareness, funding, and policy change in the rare disease landscape.

I would now like to take the opportunity to welcome our vibrant panel today.

Yamina Hsaini:
My name is Yamina and I am from France. I suffer with severe gastroparesis and other conditions that are often misunderstood, so I felt lonely and faced medical mistreatment for many years. That is why I turned to social media and created "Yamina's life", where I share my life experiences to raise awareness, change mindsets, and break the stigma surrounding rare disease. Through my activism, I was honored by EURORDIS with the 2025 Social Media Award. I am here today because I believe in the power of using your voice to bring about change, and I am grateful to be a part of this important discussion.

"I think that those of us who are able to tell our story should do so, and make sure that the stories of those who cannot are shared in some way."

Pablo Ramirez Uribe:
I live with a condition called APS Type 1, or autoimmune polyglandular syndrome type 1. It is an ultra-rare condition, which affects one in every two to three million people. My genes really decided to take the whole rare disease thing to the nth degree. When I say that I live with it, I really do mean that in every sense of the word. First off, I am very lucky to even be alive, and luck truly played a big role. When I was three years old, a Colombian doctor happened to read a study on my condition that just matched up perfectly and I received proper treatment early on. My parents and my sister have basically given up everything to make sure that I have the best care. We have moved to different places, even to the United States, 15 years ago, to be right by the NIH. If the universe allows you to live and throws luck on your lap, you should in return give that back. I feel like there is a moral responsibility for those of us who have made it. So, from using my multilingual background and working on writing to create nationally shared op-eds in Colombia, to learning at far too young an age (I was a very lonely child, I guess) how to use technology and social media, I created a YouTube channel and have been able to make some topics trend on Twitter. I also competed in speech and debate in college, where I gave a speech about rare diseases. And regardless of whether it led to being named one of Colombia's top 20 leaders of 2018, or being part of Rare Disease International's youth leadership program right now, more than anything, I keep going back to that living theme. Our Nobel Prize in literature winner, Gabriel García Márquez, called his autobiography, Vivir para Contarla, which translates to Live to Tell the Tale. I think that those of us who are able to tell our story should do so, and make sure that the stories of those who cannot are shared in some way.

Shandra Trantham:
I am a young adult in the rare disease community. I am living with Friedreich Ataxia, or FA. It is a rare neurodegenerative disease that I was diagnosed with when I was 12. I am a recent PhD graduate from the University of Florida, where I developed a gene replacement therapy for a very rare neurological disease. Being a patient and a researcher, I began to notice huge gaps in communication and became really passionate about bridging those worlds. I also learned in time that similar communication gaps exist between legislators and regulators, and patients and scientists. So I dedicated my time after school to learning all that I could to be able to actually do my part in breaking down those barriers. I am a proud ambassador of the Friedreich's Ataxia Research Alliance, and I am also a member of a young adult-only rare disease advocacy group called Young Adult Rare Representatives (YARR).

Richard Horgan:
I am the founder and CEO of a nonprofit biotech organization called Cure Rare Disease. We are advancing novel modalities to treat diseases of the muscle and brain. We are targeting diseases that larger pharmaceutical groups have traditionally left behind due to lack of prevalence, so the ultra-rare diseases, and figuring out a model by which the development of these therapeutics can one day become sustainable.

Erika Berg (*host*):
I am going to put my first question to Shandra. Your work spans both scientific research and advocacy, which are two worlds that do not often intersect. How do you see young scientists like yourself using digital tools and social media to make science more accessible and connected to the patient communities that the science can hopefully someday benefit?

> ***"It is important, as scientists, that we make every effort to share data and knowledge in layman's terms. I actually think that if you cannot explain something simply, then you probably do not fully understand it. It takes full understanding to actually be able to then put it into easier terms."***

Shandra Trantham:
As a patient myself, I think it is really important to emphasize that it is critical that we make scientific information accessible to everyone. I remember being 13 and trying to read about what was happening to my body in medical journals. Obviously, I was 13, so I had to look up every other word and it was really hard. That is why, as time has gone by and I have gotten older and learned more about science, I have used that knowledge to go back and help my patient community understand the latest news and research and things like that. Rare disease or not, we all want to understand what is happening to us. It is important, as scientists, that we make every effort to share data and knowledge in layman's terms. I actually think that if you cannot explain something simply, then you probably do not fully understand it. It takes full understanding to actually be able to then put it into easier terms.

As a scientist, I have been in plenty of spaces where others in the room had never met a patient with the disease that they were researching. It is kind of crazy. When I was in the lab, some of the people there were working on my disease and they had never met a patient before me. I have seen similar things happen with legislators. They have the power to make decisions that they do not personally know the implications of. So I have really come to realize how important it is for us as patients to share stories with these people to actually affect change. As scientists, we should really not be afraid to actually go into patient spaces, attend patient events, and things like that, so we can meet the people that we are trying to help and listen to their stories.

Erika Berg (*host*):
Pablo, you talk about telling stories and you have used storytelling, YouTube, social media, and other channels to create impact across borders and across the globe. What role do you think language, culture and accessibility play in shaping inclusive rare disease communities online, and how can digital platforms do better in that space?

> ***"I think all of us here are storytellers in that way, whether we are researchers, scientists, or advocates. Because of that, there are storytelling contours that all of us in the rare disease world can immediately relate to. A rare disease patient tells you about how a doctor did not believe them or how they were not accommodated at work or at school, and instantly, we understand. "***

Pablo Ramirez Uribe:
So, to preempt the response to that question, I am going to go off on something that may seem like a tangent, but to quote my friend William Shakespeare, "There is method to this madness." Because when we talk about storytelling, I am somebody who very deeply believes that we human beings think in stories. It is almost like a common language that we all understand instinctively. You hit us with a beginning, middle and end, and we follow you. You tell us what somebody wants and the obstacles that they are going to face, and you have got yourself a plot that is going forward. And stories, honestly, can be anything. Richard Feynman, one of my absolute heroes, did an interview in which he talks about the process that makes fire, and not only does he frame it as this story, but he even gives carbon and oxygen desires, and I love that. I think all of us here are storytellers in that way, whether we are researchers, scientists, or advocates. Because of that, there are storytelling contours that all of us in the rare disease world can immediately relate to. A rare disease patient tells you about how a doctor did not believe them or how they were not accommodated at work or at school, and instantly, we understand. Similarly, somebody might tell you about their diagnostic odyssey, or how they feel they must live their life as a rare disease patient, caregiver or advocate to make sure that somebody with the condition that they are related to does not have to deal with so much later on. All these things are a common grammar that if we hear them, we relate to them in the rare disease world. So when it comes to writing in different languages, it is not as hard in that sense. For example, in Yamina's videos, she is able to put the subtitles in French and in English. Along with the videos, you instantly realize the visceral reality of what she is talking about, which I think you get in a lot of other examples.

In terms of culture and accessibility, we live in a time in which so much is available to us online, and not just for research purposes. In 2017, when I did that speech about rare diseases, I started by talking about Julia Vitarello, the mother of this beautiful six-year-old girl called Mila who had a condition called Batten disease. I learned just a few days ago that Richard has been working with the scientists who helped Julia develop the treatment for her daughter. I think it is called Milasen. Even though her daughter passed away, she was able to do this remarkable thing. So you go from researching a story in the Denver Post in 2017, to being at this event a few days ago and finding out that there is another connection, not just to the story itself, but to the person who played an integral role in it, and who is now related to Richard's cause as well.

"The big thing for me is finding ways to support people in the rare disease community who have never been able to speak. With the technology we have nowadays we are able to do that, but how do we show them how to maximize the use of each platform?"

I love what Shandra said about making information accessible to everyone. I think that is the great thing about the tools that we have now, but where do we go from there? Like you said, I believe that as we can rely on this common storytelling language and grammar, we can fit our stories within it and we can use the tools that we have around us. These may include visual tools, like YouTube, or image-based ones, like Instagram. You also have Twitter, Threads, and Bluesky to write small posts, or Medium and others to write longer ones. The big thing for me is finding ways to support people in the rare disease community who have never been able to speak. With the technology we have nowadays we are able to do that, but how do we show them how to maximize the use of each platform? How do we show them what YouTube can do versus TikTok, even though they are both video-based? How can we show them the fact that, as I have learned, Twitter is still used overwhelmingly in Colombia, so I use that one in Spanish a lot, but other ones like Bluesky and Threads are used more in English instead. How do we allow rare disease patients and others around them to realize that we have tools that can help them translate text, with nuance, into other languages? How can we let them know that they can look up videos, interviews, articles, and books, and that because of the multicultural world we live in, it is now much easier to relate to others regardless of cultural background? I think that if we approach it in that way, all the things we have, that are helping us create new senses of connection and community through shared resources and stories, can give people whose voices we have never heard before a chance to amplify them. Then we can come in and say, "All right, how do we show them what tools are available and how to use them in the best way?"

Erika Berg (*host*):
Yamina, you have built a powerful platform rooted in your personal storytelling and your lived experience. What have you found to be the most effective ways to foster authentic connection online in a world of curated content? And how can digital spaces become safer and more empowering for young people with rare conditions?

"We can make digital spaces safer for young people by creating truly inclusive content. For example, we should focus on normalizing life with rare conditions and help others understand what it is like to live with them. By doing this, we create spaces where people are more likely to support one another. Because the more people understand, the less they judge."

Yamina Hsaini:
I believe the best way to build an authentic connection online is by showing up exactly as we are, not as society expects us to be, but as our most honest version. For example, I share my daily life on social media, including my pain and vulnerable moments, but also my victories and my achievements, despite living with chronic illnesses. By opening up about my journey, people often tell me they can really relate to me. Speaking truthfully and sharing the challenges that I face helps create and maintain an authentic community, because people connect with what they can feel and with real stories. I think we can make digital spaces safer for young people by creating truly inclusive content. For example, we should focus on normalizing life with rare conditions and help others understand what it is like to live with them. By doing this, we create spaces where people are more likely to support one another. Because the more people understand, the less they judge. Step by step, we can create an environment where young people feel safe and can show up without fear.

Erika Berg (*host*):
Richard, your journey with Cure Rare Disease is rooted in your personal story and has led to groundbreaking work in bespoke genetic therapies. How do you see young people and grassroots digital movements accelerating drug development and reshaping the future of rare disease research and biotechnology?

Richard Horgan:
I think when we look at advocacy groups, non-profits, and family foundations, I would argue that it is not so much about accelerating certain groups of research, but rather about enabling them. So, for instance, of the more than 10,000 rare diseases impacting over 10% of the United States population, many of them are just far too rare to attract commercial drug development interests because there is no return on investment (ROI) on treating those diseases. That is not a judgment, it is really just an objective statement, with regard to how diseases are treated. These groups that you are seeing coalesce and bubble up to the surface, such as the work that Julia or others have done, or the work that we are doing, are trying to develop an approach that may be sufficient to develop therapeutics to treat these diseases. For instance, if you have a disease that affects only about 100 Americans, it is very unlikely to ever attract commercial drug development interests. However, in such cases our approach is to partner with academic groups that develop early stage versions of potential gene replacement therapies. Then our staff, comprised of translational experts, toxicologists, pharmacologists, and regulatory and manufacturing experts, take that early-stage drug and usher it through the development cycle, ultimately leading to an investigational new drug (IND) submission and the start of an initial clinical trial. Where we go from there is still the unanswered question. How do we take a drug that shows benefit, that may even be approved, and develop it so that it can be administered to patients who are impacted by the disease? I think that part remains unclear. From a technology perspective, we are in good shape. Can it get better? Absolutely. Will it get better? Undoubtedly. But can we treat certain diseases from a biological and physical standpoint? I would say that, for the most part, the tools and technologies to do so already exist to do exactly that. What lacks is the societal mechanism for funding and paying for this work. The funding component involves figuring out how to

gather the millions of dollars necessary to take a drug from the initial academic bench all the way to the patient's bedside, and that is no easy task, especially nowadays. If there is a safety and efficacy profile that is acceptable to the regulators, which ultimately results in that drug being approved or perhaps even staying in perpetual IND, how do we ensure that patients can access that drug? These are the mechanisms that are unsolved today, but the way I like to think about it is at least we are not trying to change the laws of physics and biology. We are trying to modify the social fabric, and I would like to think that may be a little bit easier. The jury is still out. We will see, but that is where we are today.

Erika Berg (*host*):

Cure Rare Disease is a nonprofit. Usually when you think about biopharma, a nonprofit is not necessarily what comes to mind. Could you expand on that a little and talk about how you operate and what it means to be a nonprofit in this space?

> ***"If you looked maybe five or even ten years ago, the number of nonprofits driving drug development activities was effectively zero. Historically, nonprofits took the position of raising early dollars and then giving them to academia, or perhaps a small biotech, which would use them to develop a drug."***

Richard Horgan:

If you looked maybe five or even ten years ago, the number of nonprofits driving drug development activities was effectively zero. Historically, nonprofits took the position of raising early dollars and then giving them to academia, or perhaps a small biotech, which would use them to develop a drug. Until recently, nonprofits have not historically taken the lead and acted as the sponsor in developing drugs. Now fast forward to 2017 and 2018 with the advancement of efforts like Dr. Yu's work at Boston Children's Hospital, Dr. Neil Schneider's work at Columbia on rare forms of amyotrophic lateral sclerosis (ALS), and our work with the advancement of a personalized genome editor. Over the last five years, as these early-stage cases have come to the public's attention and have been published and discussed, they have opened up an opportunity for other groups that have historically not spearheaded drug development efforts to say, "Wait, actually we think we can do this."

So for us, as a nonprofit, there are very few, if any, investor dollars going towards funding the diseases that we work on, which is okay. How a lot of nonprofits function nowadays is through grassroots donations, NIH grants, state grants, and major philanthropic gifts. These, I would say, are unproven so far and are not sustainable. What becomes sustainable is how we can get drugs approved, reimbursed, and then reinvest those capital dollars back into new programs. So, it is a bit of a long-winded answer to your question, but I believe this new generation of decentralized drug development will especially benefit ultra-rare diseases that have historically been neglected and forgotten.

Erika Berg (*host*):

Shandra, you were discussing earlier how you communicate research with the rare disease community. How does your lived experience impact that communication compared to a scientist working in that area without any? I was wondering if you could talk about that and what role technology or other types of platforms have played in that communication.

> ***"I think technology has really helped the rare disease communities come together. We may be rare, but there are so many of us all around the world. Even though we all may have a different disease, we all share the same commonality of having a rare disease. We all share some similar struggles in getting people to understand our story and wanting to create meaningful change."***

Shandra Trantham:

Even as a scientist now, I can still remember what it was like when I did not understand everything. I was trying to understand, but it was honestly like hearing a foreign language and you do not know enough to even ask questions. I think that in order to effectively communicate, you have to put yourself in that position. Since I have been in that position in the past, I am able to think about what the news is and what I need to communicate and then how to frame it in a way that younger me would have understood. People in my community want to be informed, but do not know how to ask. So I think about all those things when I am trying to get information across. And it is not just speaking about information or writing about it, but you can also do things like videos that help share the impact or tying your personal stories into it so that people can better understand what you are speaking about and why it matters to pay attention. I think technology has really helped the rare disease communities come together. We may be rare, but there are so many of us all around the world. Even though we all may have a different disease, we all share the same commonality of having a rare disease. We all share some similar struggles in getting people to understand our story and wanting to create meaningful change.

I would say that I really began to notice the power of my own patient voice a few years ago when I came across this virtual education program for young adults in the U.S. that have a rare disease. It is called the YARR Leadership Academy and in this program, I met other patients like me who are using their stories to impact things like legislation, policy, and research funding. I formed a strong bond with my classmates who were there with me and who equally live with a rare disease. It is really empowering to use my voice to make a difference and to watch other young people use theirs. I do also want to echo what Richard was saying. I think that a lot of the problems that do exist are linked to money as it takes money to get treatments developed and approved. There are legislative mechanisms that help to bridge that gap and help push rare disease drug development forward. I think it is really important that programs like that continue to exist. I know that currently, one of these programs has not been renewed and is on pause. That could really impact rare disease program development and sharing our story can help bring that back and help create other methods of driving research and treatment development forward.

Erika Berg (*host*):
What did you learn through your education program that might help others tell their story in order to reach a wider audience and make cases that regulators or legislative bodies might listen to?

Shandra Trantham:
I would definitely say that the most important thing is tying your story to whatever you are saying so that people can really understand why it matters. The program that I was speaking about before, which has currently not been renewed, is the Pediatric Rare Disease Priority Review Voucher (PRV). This is a program that incentivizes rare disease drug development, because when a pharmaceutical company develops a treatment for a rare disease and if it makes it to FDA approval, they are granted a voucher. This does not cost the government any money, but the voucher is incredibly valuable because it actually allows for an accelerated review timeline for a drug, if you turn in this voucher. So companies will want to buy that for hundreds of millions of dollars. An example of using my story is that I have met with legislators, shared a little about this program, and explained that for a long time there were no treatments for my disease. While the fact that it takes so long to develop and review a drug through the FDA is, obviously, important for safety, as a patient it is hard to sit and wait for things to happen. Programs like this really incentivize companies to get involved in developing drugs for rare diseases, projects they might not otherwise pursue because they do not see a clear financial benefit to developing drugs for rare diseases with a limited patient population. So it is really important for this program to exist and I have met with legislators and shared my personal story to help better explain this so that people are able to have a better idea of why things like this matter. It can sound very complicated, but when you actually explain it in the context of your story it makes it a lot easier to understand and for people to see the impact.

Erika Berg (*host*):
Pablo, you have had a huge impact using various strategies and I am wondering if you could talk about the ones you have found to be most effective for increasing awareness, sustained engagement and impact. What are some tricks we could learn?

"Figure out the answer to the question, "Why should they care about it?" Because unfortunately, simply saying they should care because it is human to care is often not enough."

Pablo Ramirez Uribe:
I was actually talking about this with someone recently. We were in one of the groups from the Youth Leadership Program and we were talking about social media and the digital elements, and somebody asked, "I have only very recently started feeling comfortable sharing my story. How did each of you do it?" I basically said that I did it out of spite, and I will explain what I mean. I say this not about my immediate family, like my parents and my sister, but about other people in that orbit who would talk about the condition in whispers, as if it was some sort of taboo, yet also gossip about it. It still happens with one of my cousins. Her son also has a rare disease and I can see how these individuals behave in the same way. Very early on, I decided that I was going to talk about this at all times and that it was going to be out there in the public. I wanted to avoid the cognitive dissonance of people in my family being proud of me for who I was, because of course they would talk about that, yet at the same time not want to talk about the rare disease that was a part of it, which would create a disconnect.

I love what Yamina said about showing people how things are and not how they expect them to be, so that the veil falls down and the reality is very present. I think the biggest tool that I have found is telling things as they are. Obviously within a level of not going to a dark place where you are destroying yourself to use your own personal story and not practicing self-care, but ask yourself what are the things that you think need to be out there so that people care? As Richard mentioned, when we are talking about different conditions and you want to convince people to care about them, it is not an easy thing to do. For example, with a medication that is only for a few hundred people. As an English teacher, I would tell my students when we write persuasive essays that you need to have a "So what?" question. Tell me why I should care about the topic you are writing about. Does it connect to me personally? Is it going to affect the economy in some way? Is it going to affect future generations? And when you find that little through line that will reach a different audience member, you have succeeded. That is how you get them to understand that it actually does impact them and they want to learn more. I feel like I am still stumbling backwards into using social media and writing narratives, but there is always something new that you learn when testing things out. I would suggest that you find a way to boil down one part of your story to the basic essentials: beginning, middle, and end. Ask yourself what do you need right now? What is blocking you? Choose one avenue or one audience, whether you are a middle school or high school student talking to your classmates to celebrate Rare Disease Day, or you are a researcher or executive wanting to see if you can reach your audience. Figure out the answer to the question, "Why should they care about it?" Because unfortunately, simply saying they should care because it is human to care is often not enough.

So, find that, "So what?" which is very specific to them, and then just test out different methods. If nothing else, they are changing so fast that I think a lot of us might not even know in a few years from now what the tools are that work for some of these platforms. So if we at least start working now, while being afraid of failing and making mistakes (which is good, because it means that it is important work and we care about it), as technologies develop and the landscape of the rare disease world changes, we will already have some experience. This way, we can adapt as opposed to starting from scratch and having to learn how to do something while also navigating the rare disease world. In conclusion, I would say: find that "So what?", learn how to tell your story, and test out different strategies, because things are changing so fast and we will be the trailblazers.

Erika Berg (*host*):
Are any of you using artificial intelligence (AI) to help tell your story or in creating those connections?

Pablo Ramirez Uribe:
I am an early adopter of all types of technology, and as an English teacher who knew that students were going to start using it to write their own essays, I remember just going for it and trying it out. I tried 30 or 40 different prompts from "write a five-paragraph essay" to "write a passive-aggressive email", which I sent to two of my colleagues, who are also my

best friends. I was trying all these different things to see how this tool works and to understand its limits. Bless the heart of my students, they thought I was born yesterday and we would not catch it, but there are obvious things that artificial intelligence and generative AI cannot do. So what could it do? It could take the text or the transcript from a YouTube video in another language and translate it for us, and then that is where we add our little bit of flavor and nuance. It can help, as I know a lot of researchers are doing, with collating data about different symptoms and seeing what they reveal. I met somebody at the World Orphan Drug Congress recently who is trying to collect data for different conditions using established registries. They are using registries to collate all that data and see what symptoms are mentioned most often. That way, whenever physicians test for something, the model can use what it has already learned to support the diagnosis. We can see how effective it is at using the information it takes in to potentially diagnose someone sooner rather than later. It is a very exciting and, at the same time, terrifying tool, and I can see so much potential for it in the rare disease world.

Erika Berg (*host*):
Anyone have anything to add from the research side?

Richard Horgan:
We do use AI in a slightly different sense. We are using AI to optimize the sequences of the drugs we are developing. There are known areas where certain DNA sequences, made up of the nucleotides A, T, C, and G, elicit an immune response. What we do is we feed our sequences into an AI machine learning platform, and then that is able to generate what we call optimized sequences that are less likely to elicit an immune response. In the case of the drugs we are developing, they are delivered with a virus, called adeno-associated virus (AAV), which is an immunogenic virus. Clinically, patients have their immune systems suppressed and T and B cells interrupted so as not to elicit an immune response. In leveraging AI, our hope is that we can further improve safety. So it is very novel. The jury is still out on its efficacy, but we will see.

Erika Berg (*host*):
Yamina, you have a deeply personal and honest social media presence. Can you share with us your biggest challenge and your greatest reward in digital storytelling? And how do you keep your community grounded in a sense of hope and resilience in something that can sometimes have dark aspects?

> ***"Even when there were negative people, it never stopped me from advocating because it only reminded me of how important it is to change the way people think. My biggest reward is the community I have built and the safe space I have created, and the way I have turned my pain into my strength."***

Yamina Hsaini:
My biggest challenge has been exposing my personal life while raising awareness about illnesses. This is because it means opening up completely to a large audience, knowing that I am talking about complex and often misunderstood topics that many people have never heard of, which can often lead to confusion or judgment. Even when there were negative people, it never stopped me from advocating because it only reminded me of how important it is to change the way people think. My biggest reward is the community I have built and the safe space I have created, and the way I have turned my pain into my strength. When all the medical injustice started in my life, I did not know anyone going through something similar, but by sharing my experiences, I help others avoid the same loneliness by giving them the kind of hope I wish I had back then. I have inspired many people to accept themselves and fight to be understood. All of this keeps my community grounded in hope and resilience by accepting the struggle and growing through it together with truth and support.

Erika Berg (*host*):
Shandra, we often hear about the isolating nature of rare disease. How has technology helped you feel less alone and can you talk about any meaningful connections you have built through digital spaces?

Shandra Trantham:
I can really speak to this one. As I mentioned, I was diagnosed with FA when I was 12. At the time, I was in middle school, and obviously, I was very ashamed of being different from other people. I actually did not even tell anyone about my diagnosis until I was in college. The symptoms were obvious to other people, but I always just played it off as some other excuse, and I was really afraid to tell people what was really going on. In college, I finally started to tell some people around me that I was really good friends with, and their response of being okay with my big secret made me feel better about reaching out to other people with FA. Obviously, with it being a rare disease, I did that online. That has really changed my life and the trajectory of everything that I am doing. I joined something called the Patient Ambassador Program at the Friedreich's Ataxia Research Alliance. In that program, we meet monthly and there are usually about 50 people with FA on the call, coming from all across the country and the world. We meet monthly to connect with each other, learn new things that are going on, and find ways that we can help make a difference.

Through connecting in the Ambassador Program, I have met some of my best friends that I am very close with now. I actually lived with another person with FA. He is my best friend, and we lived together for two years, and now we are neighbors. So, it has really made a difference. Being able to connect online when you have a rare disease means you are actually able to have a community, which you may not have had otherwise due to geographic limitations.

> ***"Technology, especially social media, became the only place where I could stay connected and finally feel less alone by finding people who understand me. I just had to log in to share things I used to keep to myself. Suddenly I felt surrounded."***

Yamina Hsaini:
Personally, I was isolated at a time in my life when most young people are building a social life. Technology, especially social media, became the only place where I could stay connected and finally feel less

alone by finding people who understand me. I just had to log in to share things I used to keep to myself. Suddenly I felt surrounded. The most meaningful connections I have built were the people who supported me like I was someone close. For example, when I could not afford treatment, they helped me. When I shared my struggles, they offered solutions, advice and care. For me, digital spaces have become much more than a virtual place. They are where I found support and connection, and they literally saved my life.

Erika Berg (*host*):
Richard, I wanted to ask you about access, as the money question keeps coming up. How can we help to reduce these disparities in access and make sure that technology does not increase those gaps rather than shrink them? We are developing these amazing new research technologies, but how do we make sure that people can still access those tools?

> ***"Simplifying the reimbursement process for these ultra-rare diseases, which have limited (if any) commercial opportunities, could greatly improve accessibility."***

Richard Horgan:
I think there are a couple ways. The first is the point Shandra made about the PRV. I think for the work that we do, it is essential. It is not something that is costing the taxpayers millions of dollars. These are secondary market transactions that are not putting a huge burden on the American taxpayer, but they are hugely catalytic. I think the second way is with regard to improved flexibility within regulatory decision-making and policy. We have talked to state plans, federal plans and CEOs of private plans to get a good understanding of how they all think about paying for drugs. The short answer is that drugs are paid for once they have achieved commercial approval. This is done by obtaining a Biologics License Application (BLA) or a New Drug Application (NDA) after having gone through clinical trials. So those two things are generally joined at the hip.

For ultra-rare populations, we are never going to be in a universe where we can have 100 or 200 patients in a study; by the nature of ultra rare diseases, that will not happen. However, I strongly believe that with regard to increased flexibility within regulatory approvals, if we can show meaningful benefit against some sort of established baseline, whether the individual is their own control or there is a natural history study published for a disease, we could potentially see the commercial approval of a drug on the back of two, three, or four clinical trial patients. This may sound somewhat new, but when we are dealing with populations that are 30 to 50 patients, treating 10 to 25% of the population is meaningful. Such data should be sufficient to signal whether the drug is effective and whether it is safe. This has to be partnered with long-term follow-up, as it cannot only be a one year period and then it is over. We need to have that post-market surveillance activity going on. So I think that increased flexibility in drug approvals then allows insurance to pay for those drugs, which in turn improves accessibility. Around half of Duchenne muscular dystrophy patients do have Medicaid as well as private insurance, in some cases. Simplifying the reimbursement process for these ultra-rare diseases, which have limited (if any) commercial opportunities, could greatly improve accessibility. This, paired with the PRV, offers us the incentive to get this work done on a smaller scale. I think the challenge with traditional drug investment, from a venture perspective, is that we are looking for a one to 100-fold return on our investment. I would encourage venture groups to look at this differently: if we can create a framework where a drug for 20 people can get a commercial approval on the back of a clinical trial of two, three, or four patients, then we can make a larger number of smaller bets, and still achieve returns of 100 times our investment. The returns are just spread over a larger portfolio.

Erika Berg (*host*):
Looking ahead, what do you think the rare disease community will look like 10 years from now? What are you most excited about and how do you hope that you could potentially contribute to that vision?

Pablo Ramirez Uribe:
I was racking my brain trying to figure out what I think it is going to look like. While I cannot really answer that question, I think that we will have more people alive and connected. I think we will have a lot more family members that will see that person live and flourish, whether it is through researching treatments or the conditions themselves, or being able to share that message and speak our truth to the world. Hearing from everyone is the most exciting part and I feel very lucky. You do not choose to be born into this world, but we are in a moment in which I can say to myself, "I am very lucky to be able to share spaces like these." All I know is that I am excited because there is going to be a lot of work that needs to be done. But man, it is going to be incredibly fulfilling 10 years from now.

Yamina Hsaini:
I think in 10 years, the rare disease community will be a space where young people with chronic and rare conditions feel included from the beginning, not after years of silence or medical injustice. I hope to contribute to that future by continuing to raise awareness through storytelling and by creating new kinds of spaces to share my experience.

Richard Horgan:
I think the next decade of rare disease achievements will pragmatically look like more safe and effective drugs on the market and an ability to pay for them. Over the next few years, with these three or four million dollar gene therapies, we will see how that works. I anticipate seeing an increase in value-based reimbursement. We cannot just dole out four million dollars for a drug that may or may not work for every patient. But we will see. I think it is up to conversations like these and groups, in a broader sense, to keep pushing that edge forward into the great unknown. I am hoping for the best.

> ***"I would love to see people with rare diseases working not only in science, but also in the government and at institutions like the FDA, so that our voices can be heard on a larger scale."***

Shandra Trantham:
I think the future is going to look like a lot more patients with rare diseases in these spaces where we previously had no presence. I am now a scientist with a rare disease and my best friend, who I mentioned earlier, was also in my graduate program. So that makes two of us in science at this one school who have a rare disease (that I know of). I have also been hearing about lots of other people that I have connected with online that are in science and have rare diseases. I think that is really great. I hope that we will continue to occupy other spaces too. I would love to see people with rare diseases working not only in science, but also in the government and at institutions like the FDA, so that our voices can be heard on a larger scale.

Book # 12.27

Have your say.

Webinars:

Podcasts:

Books:

Fondation Ipsen
70, rue Balard
75015 Paris
France
www.fondation-ipsen.org

Contact:
fondation@ipsen.com

Fondation Ipsen operates under the aegis of Fondation de France – www.fondation-ipsen.org
Editing: Natasha Barr – www.caretently.com
Graphic design: Céline Colombier-Maffre
Legal deposit: October 2025