# The Rare Disease Gazette

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

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# BUILDING A GLOBAL COMMUNITY

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# Editorial

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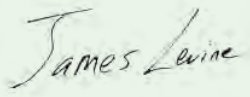
Rare Disease Detection: Rare But Not Alone

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.




## DON'T MISS!

### The Conversation

Erika Berg, Ph.D, hosts a conversation with world's experts about rare disease: Building a global community for rare disease

# The Conversation

**Experts of the month**

**Benjamin Forred, M.B.A.**
*(ZebraSite Studios, South Dakota, United States)*
**Roberto Giugliani, M.D., Ph.D.**
*(Federal University of Rio Grande do Sul, Porto Alegre, Brazil)*
**Eda Selebatso, Ph.D.**
*(Botswana Organisation for Rare Diseases, Gaborone, Botswana)*
**Rachel Smith, B.A.**
*(Parexel, United Kingdom)*

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

***"Collaborating at an international level has brought us tremendous growth. It has also given us a lot to work with, because once you connect with people that have gone before you, the speed at which you progress on certain issues is much faster. You are already ahead because you are not starting from zero."***

**– Eda Selebatso**

**Erika Berg (host):**
*Rare diseases affect millions of people worldwide, yet patients and families continue to face unequal access to diagnosis, care, and emerging therapies. Strengthening a global rare disease community by uniting researchers, clinicians, policymakers, patient advocates, and data leaders is essential to reducing these disparities and accelerating meaningful progress. Today we will be exploring how international collaboration can advance rare disease research, improve care pathways, and address long-standing inequities across regions. I would now like to take the opportunity to welcome our panel today.*

**Eda Selebatso:**
My name is Eda Selebatso. I am from Botswana and I am also based in Botswana. I am a mother of two children with rare diseases, the oldest with an undiagnosed kidney disease, who had to undergo a kidney transplant at the age of 6 and who is now 17 years old, and a daughter with Morquio A (also known as mucopolysaccharidosis type 4A). We received her diagnosis when she was 3 years old, after sending her samples to NHS Manchester in the UK. That basically sets the stage for where we live and what access looks like for us. Therefore, due to my personal challenges, I established an organization for rare diseases to make sure that we could reach out to other families struggling with the same issues that we have had to grapple with. I have been in this space for over 10 years. I am not only doing work in my own country; I also have responsibilities globally where we collaborate with other international organizations focused on rare diseases and I serve on different boards.

**Rachel Smith:**
I think this proves how small the world is. I am actually in the UK and I am just south of NHS Manchester, so I am very familiar with the rare disease work that they do. My name is Rachel Smith and I am currently working in the clinical research space at Parexel, as the Vice President and Head of the Rare and Genetic Disease Group. I work in global drug development, and I do truly mean global. I have worked very closely with the Middle East, Africa, India, China and most of the APAC countries, as well as Europe, North America and Latin America over the course of my career that now spans two decades. I am also a rare patient myself. I have a rare blood condition and it took me quite a while to get diagnosed, despite working in this field and despite working with advocacy groups. I was familiar with the patient journey and knew the signs and yet I struggled to identify them in myself. I am based in the UK, so you would think that it would be an easier diagnostic path for me. It unfortunately was not, so I am really thrilled to speak about the global approach to rare diseases today.

**Roberto Giugliani:**
I am Roberto Giugliani. I am a medical geneticist based in Porto Alegre in the south of Brazil and I am a professor at the Federal University of Rio Grande do Sul. I also work in the University hospital as a medical geneticist and I am a consultant for Dasa Genomics. I am now the director of a new rare disease center, which is called Casa dos Raros (House of Rares in English), which was established a couple of years ago and is developing some innovative actions in the field of rare disease.

**Benjamin Forred:**
My name is Ben Forred. My background is in cell and molecular biology. I spent about 15 years as a researcher working at a large academic hospital system in the United States and I have done rare disease research at the bench, but I have also helped lead a team of clinical researchers on our genetics, genomics and rare disease portfolio. I also directed the Coordination of Rare Diseases at Sanford (CoRDS) Rare Disease Patient Registry for about 10 years.

**Erika Berg (host):**
*I am going to put this first question to all of you. Across the entire range of rare*

*disease research and care, which parts benefit most from having a truly global and inclusive approach?*

> ***"When you are somebody who is located in a place like Botswana, and this applies to the whole continent of Africa, resources are limited and you find that other countries with resources are way ahead when it comes to rare disease research. However, this does not mean that the resource-limited countries have nothing to offer."***

**Eda Selebatso:**
When looking at research and even care, I would say that they would both benefit from a truly global and inclusive approach. I will start with research. When you are somebody who is located in a place like Botswana, and this applies to the whole continent of Africa, resources are limited and you find that other countries with resources are way ahead when it comes to rare disease research. However, this does not mean that the resource-limited countries have nothing to offer. You soon realize that research informed by local findings, misses insights that could be learned from places like Africa, which has a large population and could play an important role for things like clinical trials. Even though little is currently being done, there is a huge opportunity to gather a great deal of valuable information. This is especially true because there are lots of differences in our populations. Our cultures, our environments, and our genetics are different. Therefore, there is value in looking at other places even though there might not be resources available.

When it comes to care, there is also an opportunity to collaborate and exchange information. For countries without much information on rare diseases, there is no need to start from the ground up. There is already information out there that could be shared and could benefit the patients. There could also be a sharing of the rare disease specialists, who would benefit by taking care of patients that they would not naturally encounter in their usual care settings through the use of technology. The countries that do not have such specialists would also benefit and that access could then be made easier.

> ***"If you are only looking at 20 patients in the US or five patients in the UK, you do not get a representative understanding of what that condition or its natural history looks like and without that, we cannot know what we need to focus on to treat it."***

**Rachel Smith:**
When it comes to rare diseases, we are talking about very small populations within each country, but together they are not. The number of rare diseases changes every day, depending on what source you look at, and that is a part of the problem because we do not know how many people are impacted by rare diseases. So, we really need to work globally. If I was going to highlight one thing, it would be data sharing. About 80%, or potentially even more than that, as we are identifying more and more rare conditions every day, are genetic in nature. So, we have all of these genetic variations and different phenotypes. If you are only looking at 20 patients in the US or five patients in the UK, you do not get a representative understanding of what that condition or its natural history looks like and without that, we cannot know what we need to focus on to treat it. A lot of these conditions are also multi-organ and multisystem diseases and so, not just affecting the skin, kidney or brain. We are really looking at global conditions. By bringing all of that data together, from Botswana and all over Africa, China, and India, and integrating it with European and American data, it will allow us to have this full understanding of what the solution looks like. We will be better able to understand how to treat the disease, which will lead to more research opportunities and the development of more drugs. For me, that is the key piece.

**Roberto Giugliani:**
I can give many examples, but I will focus on one that occurred a few weeks ago. We had a conference on undiagnosed diseases and it was organized for the first time in the Southern hemisphere in Latin America. It took place in Rio de Janeiro in Brazil. We had around 400 people from 45 countries discussing how to approach patients with an undiagnosed disease. This is because often we perform an investigation to identify the disease and to make the diagnosis, but we do not always reach a conclusion. This patient, therefore, remains without a diagnosis and without a diagnosis it is difficult to receive proper treatment and management. So, it is very important to have a diagnosis. In developed countries, and especially in the US, Canada, Europe and Japan, there are programs for undiagnosed diseases, but most lower- and middle-income countries do not have these types of programs. So, we have now established Undiagnosed Diseases Network International, which aims to disseminate this kind of approach and tries to expand the possibilities of making a diagnosis in patients with a rare disease. We had this conference and, as a result, many countries established their own undiagnosed disease program. This is an example of how international collaboration can advance the field.

> ***"As a person who lives with a rare diagnosis, I can tell you that we all start as undiagnosed and so, we know how that feels, and we also know what it feels like to get an answer, even if it is not one that you are really happy about."***

**Benjamin Forred:**
A lot has already been touched on, but I think there are two things that jump out at me in terms of global impact. One would be advances in genetic testing globally. The more genetic testing we can do, the more we can understand the human genome across all of our differences, including in race, gender and wherever we are on earth, and that will help eliminate variants of unknown significance (VUSs) and understanding more about how systems play together. The other major point is the advances that are being made in data science, where we are taking all this healthcare data from

different systems, countries and places, and then finding common models that can be used to organize that data so that it can be analyzed across all sorts of different contexts. We are now getting to a point where we can take a global approach at looking at what makes us sick. As a person who lives with a rare diagnosis, I can tell you that we all start as undiagnosed and so, we know how that feels, and we also know what it feels like to get an answer, even if it is not one that you are happy about. So, I think that the advances in data science, genetics, newborn screening, and similar things are going to have a huge global impact.

**Erika Berg (host):**
*Roberto shared a brilliant example of where international collaboration is making a difference. Does anyone else have any other examples they would like to share of a recent experience where international collaboration really demonstrated the power of what working together can do for rare disease?*

**Benjamin Forred:**
I have been working with the International Rare Diseases Research Consortium (IRDiRC) and have been on a task force for preventive medicine for the past year. About a dozen of us have been meeting regularly to put together a white paper about different approaches globally that can be taken in regard to preventive medicine for rare disease, which is an interesting thing to wrap your head around. That has been a really great task force that has come together across different continents and different backgrounds to understand that there is not really a one-size-fits-all solution. Being nimble and being flexible is going to have to be a major component of moving the needle and advancing research for rare diseases globally.

***"This year we were very fortunate to collaborate with the European Commission and EURORDIS, with an initiative called EU-X-CT, which is essentially opening up cross-border enrollment to increase access to clinical trials. This means that even if you are not located in the country where a clinical trial is being run, you can still access the clinical trial and its treatment options."***

**Rachel Smith:**
I have a couple of examples. To add to Ben's earlier point, I think there is so much going on in this space. There are so many groups that are either individually or collectively working together. So, you have the N of 1 space where there are individuals that are not usually N of 1 when we start looking closer, but they start off that way. Then we have basic examples from the clinical space. This year we were very fortunate to collaborate with the European Commission and EURORDIS, with an initiative called EU-X-CT, which is essentially opening cross-border enrollment to increase access to clinical trials. This means that even if you are not located in the country where a clinical trial is being run, you can still access the clinical trial and its treatment options. We are often working with patients with these really rare diseases and conditions and we are opening a site in Canada and in Germany and the patients are located all over the world.

We had a patient recently, a young boy, who lives in Kazakhstan and we were very fortunate to be able to link him with an investigation from a site in Germany and actually had that patient enroll in a 12-month clinical trial where he opted to stay in Germany for those 12 months. That would not have been possible if we did not have the guidelines and the regulations in place that allow for that kind of access to healthcare in the clinical trial setting. So, that guidance is really benefiting patients. That is one case in particular, but we have hundreds of these patients in clinical trials where it is not in their home country, which is amazing.

**Eda Selebatso:**
I will touch on three things that have tremendously impacted patients' lives. Diagnosis is a big one, as we do struggle with diagnosis in this part of the world, especially with rare diseases. As Ben has said, genetic testing is the most important tool for reaching a diagnosis but there are infrastructure limitations that affect access to it. So, we have collaborated with Genetic Alliance to continue how we have been collaborating globally to get a diagnosis. The difference is that we are now able to get more patients treated through the iHope program, which is spearheaded by Genetic Alliance. Without such collaborations, we would not be able to go further with a diagnosis. Secondly, in regards to treatments, our government in Botswana does not offer treatment to patients with a lot of these conditions because of the costs. Therefore, we have some patients that have been treated through charitable access programs that we are involved in. In terms of policies, a lot of our countries do not have policies that are specific to rare diseases. But we are aligned with international organizations. We are happy that the recent World Health Assembly (WHA) Resolution on Rare Diseases has been achieved and we have been a part of that process as representatives from Botswana working with other countries. So, these types of collaborations are very important because once you have something like this WHA resolution, you have a base on which to start and continue conversations and have guidance for all countries on how to approach the policy issues.

**Erika Berg (host):**
*What are the biggest gaps that you see in rare disease resources between high-income countries and the rest of the world? Is it in diagnostic testing or is there something else you are seeing?*

***"When it comes to access to treatment, I think there is an even bigger gap between high-income countries and low-income countries because the access to these expensive therapies is very limited. There are some charitable programs that contribute a little to decrease this gap. However, we still require some innovative solutions for this issue, as it will not be possible for middle-income countries to face the cost of some of these expensive therapies."***

**Roberto Giugliani:**
I think there are many large gaps and diagnosis is one example. It was already mentioned, but access to diagnostic tests is still very limited in low- and middle-income countries, and this is a major challenge. In Brazil, we are establishing a kind of collaboration where we raise money so that we can offer diagnostic tests for several diseases to countries in Latin America that do not have them available. Now, with the use of dried blood spots and urine-impregnated filter paper, we can ship these samples without ice or a cold chain, making it easier to cross borders and support this international collaboration in terms of diagnosis. It was already mentioned, but when it comes to access to treatment, I think there is an even bigger gap between high-income countries and low-income countries because the access to these expensive therapies is very limited. There are some charitable programs that contribute a little to decrease this gap. However, we still require some innovative solutions for this issue, as it will not be possible for middle-income countries to face the cost of some of these expensive therapies. There are some alternatives that we can discuss later on.

***"At the Botswana Organization for Rare Diseases (BORDIS), one of our objectives is to ensure that, wherever resources allow and opportunities exist, we work with our doctors to help build their capacity. This includes not only doctors, but other healthcare providers that offer support for the care of our patients."***

**Eda Selebatso:**
Besides what we have already mentioned, there is also the capacity gap among healthcare providers. It is a huge gap and it is very critical to how you first get diagnosed, because it is very much hinged on the doctor's index of suspicion. If someone has never encountered a rare disease, or that particular rare disease, it is going to take a long time for them to eventually prescribe the test needed for a diagnosis. At the Botswana Organization for Rare Diseases (BORDIS), one of our objectives is to ensure that, wherever resources allow and opportunities exist, we work with our doctors to help build their capacity. This includes not only doctors, but other healthcare providers that offer support for the care of our patients. We have actually hosted two trainings for African doctors in Botswana, and after those trainings, we have seen a tremendous increase in the number of lysosomal storage disorders being diagnosed. So, that is one important issue that still needs to be worked on.

**Erika Berg (host):**
*Ben, when it comes to improving capacity in countries with fewer resources, what kinds of investments in training, technology, and infrastructure can create the biggest improvements for rare disease care?*

***"Being able to have a patient receive a diagnosis is one thing, but to have a family understand what that diagnosis means is another. When you are talking about rare disease, receiving that diagnosis is too often the end of the conversation."***

**Benjamin Forred:**
That is a great question, and it might sound like a cop out, but I think it is a combination of those three different things. You need to have the infrastructure in place. If we take something like newborn screening, you need the hospital systems and the professionals to have the ability to order the tests, have them sent out, get the results sent back, and then have people there to analyze them. There is also a necessary component of that which is focused entirely on healthcare literacy. Being able to have a patient receive a diagnosis is one thing, but to have a family understand what that diagnosis means is another. When you are talking about rare disease, receiving that diagnosis is too often the end of the conversation. There is not a, "And this is what are we going to do about it" portion. Only 5% of rare diseases have any kind of therapy or approved treatment available. So, you are stuck trying to fight symptoms instead of a root cause. You need to have a strong health literacy program, with either the help of genetic counselors or social workers at the hospital, so that people can understand how this is going to impact their life and what resources are available to support them. Without the ability to test people and then effectively help them manage their lives, you are not going to have the impact that you desire.

**Erika Berg (host):**
*Rachel, we often hear about these collaborative efforts, such as IRDiRC, that are looking to help rare disease communities globally. In your experience, how are these collaborations speeding up research and improving access to care?*

***"We have some great examples from the COVID-19 pandemic where vaccine programs globally were partly funded by those higher-income countries to give access to the vaccines, when they were available, to lower- and middle-income countries. I think something like that would be really helpful for rare diseases."***

**Rachel Smith:**
The biggest thing they do, and IRDiRC is a great example of this, is they shine light on rare diseases. To follow up on Eda's point, the fact that we have the resolution recognized by the World Health Organization was absolutely huge because that was really the recognition of what we have all been saying: that rare is not rare and you do need to have a global and international approach to it. I think IRDiRC has done an amazing job along with C-Path, EURORDIS and other organizations that are either cross-country partnerships or broader collaborations aimed at improving research. So, they are improving awareness around rare diseases. If the symptoms seen together do not make sense, then it is probably a rare

condition and we should go ahead with some sort of genetic testing. So, there is a need to increase the access to those sorts of tests. Georgia is an example of a country where we work quite frequently and they have a partnership with a lab in Germany to do all of their genetic testing and that has reduced the time it takes to access that data and access those results. That is really important because they do not have the infrastructure themselves to do that testing. A lot of that has been possible due to the national and international organizations building the guidance, policy and regulation changes, and making access to those things much less difficult. I think also harmonization and guidelines are important. Yes, there will be local healthcare system and regional differences, but ultimately rare conditions as a whole have a lot of commonalities and they are really heterogeneous. We are talking about small populations in the individual countries, the delays to diagnosis, and the lack of access to research or to approved drugs. Being able to put frameworks and best practices in place allows countries like Botswana to say "There is a framework here for us to work from." They may not be able to implement everything and these pieces may not make sense for their healthcare system or they may not be able to fund them, but I think having those pieces of data is really important. The other piece for everybody is the funding. We have some great examples from the COVID-19 pandemic where vaccine programs globally were partly funded by those higher-income countries to give access to the vaccines, when they were available, to lower- and middle-income countries. I think something like that would be really helpful for rare diseases. These organizations have funding programs to look at implementing certain task forces or infrastructure in those countries or giving them access to infrastructure that already exists in another country. So, that funding is sporadic, it is there, but it is not on the global scale yet. But again all, of this has really advanced research. IRDiRC, as mentioned, is a great example because we have had over 100 therapies that have been notably accelerated as a result of that organization independently. Obviously, there were many more examples, but that gives us a very clear figure.

**Erika Berg (host):**

*Roberto, can you tell us more about the Rare Disease International (RDI)-Lancet Commission on Rare Diseases? What are your hopes for how it might shape global rare disease care?*

**Roberto Giugliani:**

The RDI-Lancet Commission on Rare Diseases was established last year, and it came after the UN resolution about making rare diseases a global health priority. We realized that it was important to organize the view of the international community about rare diseases. So, we formed a commission comprised of 27 commissioners coming from different countries, with all continents as well as high-, low- and middle-income countries represented. We are currently discussing how to present this topic of rare disease, to set the scene for the WHO.

In 2025 the World Health Assembly approved a resolution designating rare diseases as a global health priority and now the WHO will establish guidelines for the member countries about rare diseases. So, we decided to form this commission, and we formed five working groups. One is focused on the ethical and moral aspects. Another is focused on the data and metrics, because when we talk about 6000 or 8000 rare diseases, we should really look at these numbers and be able to provide evidence when we say 70% or 80% are genetic. We also have a working group on societal and healthcare systems to approach the diagnostic odyssey of these patients. There is equally a working group on clinical pathways to examine the therapeutic alternatives. Finally, there is a fifth working group on healthcare professional competency. The importance of having these healthcare professionals trained in rare diseases was already mentioned. These are the five working groups that are working hard to produce documents that I think will be very instrumental for the people working in these areas and especially for the WHO, who should issue some resolutions and guidelines in the next years about rare diseases. So, I think we are in a very important moment, and the RDI-Lancet Commission on Rare Diseases will provide some important information and evidence for these documents that should arrive in the coming years.

**Erika Berg (host):**

*When will we get these documents? I know it is a work in progress, but do you have a sense of when they might start coming out?*

**Roberto Giugliani:**

We published a very brief document earlier this year stating the foundations of this Lancet Commission. It was published in *The Lancet* in the February 2025 edition. The five working groups are currently working in parallel and I think we should have a final document for publication in early 2027, because we will take all next year to finalize our work.

**Erika Berg (host):**

*I am going to switch gears a little to talk about patient registries, which are central to rare disease progress. Ben, what advances have you seen in making these registries more globally representative? Could you maybe talk about why they are important, your role in the CoRDS Registry and how that fits into these efforts for building these registries?*

> ***"I think the biggest success that we have had is in making sure that regardless of what language a person speaks, where they live or their economic status, they are freely able to contribute and tell their story via the CoRDS registry. That is something that I personally feel really proud of."***

**Benjamin Forred:**

The first thing I will say is that in rare disease, everyone has to acknowledge that the experts in the room are the people living with the disorders. So, to start making progress anywhere, you have to let them tell their story. A registry is at its core a tool. It is a set of questions built on a collection of demographic and contact information, which allows for the longitudinal gathering of data as life goes on for rare disease patients. Across the community as a whole, you can identify things that are common. Just looking at my experience with the CoRDS Registry, I could provide many examples of how researchers, who have spent a lot of time focused on a given condition or a given organ system, were able to identify new symptoms or pathways involved

in a given rare condition because of the registry data that was collected from that community. One of the most important things that has been done to make registries more approachable internationally is the understanding that ICD-10 codes and the way we classify disorders were designed for billing purposes and using them for research purposes can be a little ham-fisted at times. So, there has been a lot of work done to try to find ways to consistently label a given disorder so that across systems you can understand that we are talking about the same thing. One of the groups that has been very influential in this area is Orphanet with their ORPHAcodes and their list of rare diseases. We are now able to assign a number to a given diagnosis, which may go by 30 names across the globe but we understand it is the same entity. We need to be able to refer to a diagnosis consistently in a research setting, so that has been a huge accomplishment.

In terms of CoRDS, I think that when it started it was about recognizing and understanding that there is no easy way for people to access research and to overcome that initial hesitation about participating. There have been enough researchers behaving badly over time to give people good reason to be a little skeptical. So, there is this layup situation, where patients can participate in research early on and we basically said that we would try to create registries for all rare conditions and understand how that works. Our biggest challenge in the beginning was finding people. It is really hard because there is no directory and rare diseases do not obey geographies or languages or races or anything else. So, you must look at the entire world. We took the approach of partnering with advocacy organizations and patient groups around the world that are focused on serving their local communities and making sure that their people have the resources they need. We might not know where everyone is, but those groups do. So, over a period of about 10 years, we partnered with 150 to 200 different advocacy groups, and we built them free registries so that from an early stage they could begin collecting structured data under an IRB protocol. This ensures that proper consent is in place, people understand what they are getting into, and that registry data can then be used down the road for any number of things. I think the biggest success that we have had is in making sure that regardless of what language a person speaks, where they live or their economic status, they are freely able to contribute and tell their story via the CoRDS registry. That is something that I personally feel really proud of.

**Erika Berg (host):**
*Building trust for these patient registries is essential. Eda, what is your experience with patient registries and how do you make sure patients, especially those in resource-limited settings, feel confident about participating in patient registries and see the potential benefits of the research that might follow?*

> ***"Trust is built at the local level. For international registries, work has to be done with patient organizations that are in the specific countries or regions, and it does not just start with a registry."***

**Eda Selebatso:**
Though we do not have any registries in Botswana and there are a limited number of country-specific registries in Africa, we do contribute as rare disease patients to international disease-specific registries, as Ben has just mentioned. Trust is built at the local level. For international registries, work must be done with patient organizations that are in the specific countries or regions, and it does not just start with a registry. What builds trust, in my experience, is that as we work with patients for things that are care-related, you interact with them and their data. The way you deal with their data builds trust. How you deliver on things that they are expecting from you builds trust. So, whenever you say, 'we are now starting registries,' that trust has already been built through the day-to-day engagements related to care and the other social aspects of their lives. Because at BORDIS we say that rare diseases are not just a health issue, they affect every aspect of a human's life.

So, that is how trust is built, from the very beginning. Then, the patient organizations are just a trusted bridge for the patients to say they can participate in the registries. I have worked with different patients that have said, "I have seen this registry that is related to my condition. Is this a good thing to do?" What we do is we sit down and talk about it and share the objectives so that they can see how they are related to their conditions, even if they might not necessarily benefit from it directly. They then start to see how it relates to their own condition and to their wider community. It is not just about contributing for a direct personal benefit, but you also look around you and consider what this information can achieve in the long-term. It is also important to outline how their data is going to be handled and the control that they have over their data. Will they be able to remove their data if needed? Who is going to use their data? Who is going to look at their data? These are the issues that are very important when we discuss registries with patients and having the patients very much involved is critical to building registries.

**Erika Berg (host):**
*Rachel, I wanted to ask you what are some of the cultural, regulatory or technical barriers that still make data sharing difficult and are you seeing solutions that are giving you hope?*

> ***"Even in high-income countries like the US, there are real problems around that trust for data sharing and where that data is going."***

**Rachel Smith:**
I just want to add to what Eda said. Trust is based on communication and transparency, and that is the foundation for everything. If we can do that correctly, it makes everything a lot easier. What then stands in our way, as you alluded to, is the variability. I think data protection regulations are there for a reason. While I am in the UK, we were part of the EU when the general data protection regulation came in, so we are protected by that. The challenges we have are that data protection regulations vary by country. In some cases they are super minimal or even non-existent and that can really erode trust because we want

to make sure that we are protecting every individual's health data, as it is extremely sensitive data. So, what we are collecting cannot be identified back to the patient. If you are considering the United States, for example, with the healthcare insurance question, we have challenges with patients not wanting to have genetic sequencing performed. This is because if the genetics come back with anything other than something related to the direct condition that the insurance company already knows about, it can affect their premiums and their coverage.

Even in high-income countries like the US, there are real problems around that trust for data sharing and where that data is going. Then you have the European Union, which in theory has a harmonized data protection regulation. However, the interpretation of that data protection regulation varies by country, and it also varies by region within that country. In some places, it can even vary by the institutional interpretation of that regulation. I will give you a great example. We had an Italian patient who wanted to take part in a clinical trial in Germany and we agreed. One of the main things that we needed to do was get that individual patient's data sent from their physician in Italy to a physician in Germany. That, in theory, should be a really straightforward endeavor. It happens all the time between referring physicians. So, why could it not happen in this setting? In this case, because the process was going to be so lengthy, the patient was going to miss out on the clinical trial if we went through these local data protection requirements. What we ended up doing was we had the physician print out this patient's notes, and as this is for an ultra-rare condition, the notes were significant. They had to print them off and the family had to physically take these paper notes all the way to Germany to give to the physician. This happened last year, when we have computer systems, electronic medical records, and all of these technological capabilities. There may sometimes be internet access problems, but I do not know many countries and regions where people do not have access to cell phones or basic technology, and there is still a major problem with sharing data between countries. So, for organizations interested in conducting research globally, sometimes it is just seen as too difficult and too complicated and it discourages them from doing that research and sharing that data. Even the European Reference Networks that were set up as a European, publicly-funded body have had problems building their own registries due to the GDPR. So, we have needed to find solutions for the patient advocacy groups so they can get around some of the barriers, but it has been really challenging.

I think that as we get into the AI world, and we are in the AI revolution right now, the utility of AI will become clearer as we understand a little bit more about digitization, data standards, and effective guardrails. That is really important as there are limited guardrails in place for the implementation of AI at the moment. As that becomes clearer, I do think people will naturally become more literate in data and understanding what happens when your data goes into a system and that will naturally help to break down some of those barriers that we see.

**Erika Berg (host):**
*Eda, could you talk about your organization, BORDIS, and give us a little background about how it got off the ground, how you built networks with other global advocacy groups and then maybe talk about what role organizations like yours play in shaping clinical trials and how you think that role is evolving?*

***"Collaborating at an international level has brought us tremendous growth. It has also given us a lot to work with, because once you connect with people that have gone before you, the speed at which you progress on certain issues is much faster. You are already ahead because you are not starting from zero."***

**Eda Selebatso:**
Establishing a patient organization is deeply rooted in the local context. So, you deal with the internal requirements within your country first and then build from there. From my experience, I had to start locally, dealing with whatever investments we needed and complying with the regulations that we needed to meet. Once that was set up, we then went out to see who we could partner with and what international organizations were there for us to affiliate with. That is when we joined RDI, IRDiRC and other international organizations. Growth does not just happen because you are affiliated with someone. You affiliate with them with your objectives in mind and knowing what gaps you want to fill. Then, once you collaborate with them, you build and grow from there. That being said, collaborating at an international level has brought us tremendous growth. It has also given us a lot to work with, because once you connect with people that have gone before you, the speed at which you progress on certain issues is much faster. You are already ahead because you are not starting from zero. So, that has been quite positive for us.

You also asked about the evolution of patient advocacy when it comes to clinical trials. In the international organizations that we are affiliated with, I am sure people must always say, "Eda is always going to bring this up," but Africa is way behind. I always ask directly, especially when the pharmaceutical industry is in the room, and I say to them one by one, "You do clinical trials, why are we not having sites in Africa? Why is Africa only recognized when someone is looking for a market to sell their products?" I get different answers, of course. But in a nutshell our advocacy and our voices are not included when it comes to clinical trials, and it is up to us to bring them to our doorsteps. Africa can handle clinical trials. Some people may doubt this but I have worked in health research. We conduct clinical trials across the continent. There are institutions that are well-equipped with people that can handle clinical trials. So, I guess it is a matter of continuing the conversation and not just talking but showing people what could be possible if they partnered with us.

**Roberto Giugliani:**
I think this is a very important point about international collaboration in clinical trials. We just had a conference, organized by IRDiRC, the European Rare Disease Research Alliance (ERDERA) and RDI, to foster international collaboration in clinical research for rare diseases. The topic of Africa was mentioned, as well as other countries that do not usually participate but have patients and have infrastructure. We really need to strengthen these links because for rare diseases it is essential to collaborate on clinical development.

Patients are dispersed around the world, so international collaboration is crucial. What was mentioned about registries is very important because one of the things that lead us to create the RDI-Lancet Commission was the need for increased visibility of rare diseases. Registries contribute to increasing this visibility and also pave the way for clinical research because when we know where the patients are this makes it easier to organize a clinical trial. So, this is something that is really very important. Registries also contribute to a better understanding of the natural history of the disease, which is essential when designing a clinical trial. We need to know about the natural history of a disease and this information can come from the registries. So, registries are important, international collaboration is important, and we should also increase the participation of countries in Africa, Latin America, and some parts of Asia in these trials.

***"Often it is the sites located outside of the US and Western Europe that are our highest performing centers. They enroll the most patients and their data quality is amazing."***

**Rachel Smith:**
So, firstly, I am going to be inviting Eda to all my meetings with the sponsors that we work with because I am consistently fighting to bring clinical trials to any region other than the United States, Canada, and Western Europe. It is an uphill battle. I think the default is always to go to those countries for various reasons. FDA requires certain amounts of data, and often we do get exclusions and exceptions for rare disease, but that is still the default. To add to Eda's point, there is a perception that the infrastructure is not as established in these countries; however, clinical trials can absolutely be performed to an extraordinarily high quality. Often it is the sites located outside of the US and Western Europe that are our highest performing centers. They enroll the most patients and their data quality is amazing. It is only once pharmaceutical or biotech sponsors start to go to those regions that they suddenly have a light bulb go off and think why have we not done this before? But it takes a lot of convincing and honestly it requires some evidence and proof points to enable them to make that jump because they have to go back to their investors and their board and they have to justify why they decided to collect data in certain countries. Obviously, when it comes to rare diseases, there are unmet needs in every country in the world. So, we can run a trial in every country in the world and there is no reason why we should not. It is just about making people feel comfortable so that they can make that leap. As I said, Eda is welcome at any of my calls that I have with sponsors because I think she has made an incredibly compelling arguments for Botswana and Africa.

**Erika Berg (host):**
*What is one change that you would love to see that would make the international rare disease research and care community more equitable, more connected, and more inclusive for everyone?*

**Benjamin Forred:**
It may be a bit pie-in-the-sky, but I would love to see more of a global approval process. People with rare diseases have it hard enough. They did not choose where they were born and so, it should not be held against them from a policy standpoint. They are all prospective trial participants and future consumers of those treatments. So,it would be great if policymakers and industry were able to look, at least for rare conditions, at how we can get this approved in a better, more global way so that it can be accessed more equitably.

**Roberto Giugliani:**
I will highlight the initiative of RDI to establish a global network of rare disease centers. Our center in Brazil, for instance, is participating in this global network. So, we have the same patient-centered approach to diagnosis, care, training and research in rare diseases. We will also partner with other centers in Africa, Asia, and Europe that do similar things. I think we will learn from each other and we will grow as a community. I think this is a very good initiative that will make a difference in the coming years.

***"If we could have a system where any patient in the world with any condition could access the right specialists and receive the right therapies, that would be a dream."***

**Rachel Smith:**
This goes back to my initial point around data sharing. It is about acknowledging that individually we are rare, but collectively the word rare disease is in itself a misnomer. It is something that is actually an inaccurate description of the community as a whole. We use cancer as a collective, when individually certain cancers can be rare diseases themselves. I think that we need to expand the international recognition that this is a global burden and requires a global focus. The resolutions are great. I want to see action on those resolutions and I want to see changes implemented at a global level rather than just regional small changes that are not really moving the needle significantly. So, again, this is a little bit more pie-in-the-sky, but if we could have a system where any patient in the world with any condition could access the right specialists and receive the right therapies, that would be a dream.

**Eda Selebatso:**
It is a known fact that countries that are resource-strained cannot, in a short time, build the brick-and-mortar infrastructures that are needed to ensure that rare diseases are well taken care of. So, when I thought about this question, what came to mind, as Roberto has already said, was networking. The technology used for networking is growing rapidly and the networks already in place at RDI could be improved and enhanced so that a doctor located anywhere in the world, provided they have access to technology, could talk to an expert specialist who could assess the patient. Like this, the patient can get the help they need without us saying we cannot build this and that. So, we need to focus more on networking and strengthening our collaborations across the globe. That is where we could approach equity at a steady, or maybe increasing, pace.

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