Charlene Son Rigby – CEO, Global Genes

Patients need a real seat at the table, and their experiences, urgency, and insights are what push this entire field forward

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Charlene Son Rigby, CEO of Global Genes, shares her insights on the rapidly evolving rare disease landscape. She highlights exciting developments such as patient-driven research and development and the future potential of platform technologies and individualized medicines. Drawing from her personal experience, Charlene also emphasizes the critical importance of ongoing support for the rare disease community. She explains how Global Genes balances its efforts between advancing research and fostering multi-stakeholder collaboration, while ensuring patients remain at the center through advocacy and support programs.

For our audience who might not be familiar with Global Genes, can you start off by walking through the organization's history and its unique role within the rare advocacy and research landscape?

Global Genes was founded 17 years ago with a simple but important goal of helping build a rare disease community. At that time, it was incredibly difficult for rare disease patients to find one another, create support networks, or connect with researchers. Global Genes became a place where patients and advocates could meet, share experiences, and start pushing the work forward in their own disease areas. A lot has changed since then. Today, the rare disease ecosystem is much more connected. The internet has made it far easier for patients and families to find each other and to link up with scientists, academic groups, and biopharma.

Global Genes still serves as a first stop for many patients who are newly diagnosed or still searching for a diagnosis. We help them find resources, understand what comes next, and begin navigating the space. But we've also expanded our focus. Today, a major part of our work is helping patients and advocates build strong, sustainable communities and giving them the tools and support they need to help drive research and progress in their disease areas.

One of the biggest shifts we've seen in the past two decades is that patients and advocates are increasingly stepping into leadership roles within their communities. They're often the ones who feel the urgency most, and they're taking action. That was certainly true in my own life. My daughter was diagnosed with a rare neurodevelopmental disorder when she was three and a half. At that time, only about 200 children worldwide were known to have this condition, though we now know that the estimated incidence rate is 1:26,000. While some of the basic science had already been explored, no one had started moving toward therapies. So our family, along with five others, created a foundation to get research off the ground. Families being the ones to initiate the early steps is something we see happening for many rare diseases, and it marks a very exciting time.

In 2023, Global Genes merged with the nonprofit data-sharing platform RARE-X, putting into place a new strategy focuses on providing tools, connections, and opportunities to meet the needs of next-generation advocacy. Why is patient-owned, structured data so critical within the rare disease space?

Before I joined Global Genes, I was originally part of the team at RARE-X, although I wasn't one of the original founders. The idea behind RARE-X connects to what I mentioned about how patient advocates are increasingly building the tools and assets needed to move therapy development forward. Many rare disease communities start with almost no structured data to describe their condition, and when data does exist, it's often scattered across different academic labs or held within biopharma. That's traditionally how registries and natural history studies have been run.

What we now understand, especially in ultra-rare diseases, is that we simply don't have enough patients to afford fragmentation. We can't have five or six separate datasets that don't talk to each other. At the same time, while patient advocates are highly motivated to build these resources, most aren't researchers, principal investigators, statisticians, or survey designers, and they shouldn't have to be. RARE-X was created to solve that gap. The goal was to give patient communities a way to quickly set up high-quality, structured data collection that helps them understand their disease and move toward clinical trial readiness. It's about giving them the

building blocks to take data registries forward with their communities.

How would you describe the breadth of this initiative today?

The RARE-X initiative has grown tremendously. We now support data collection for more than 90 rare diseases, working with about 130 patient advocacy organizations. Something I'm especially proud of is that nearly 40 percent of participants come from outside the US. Rare disease doesn't follow geographic boundaries. Knowing where patients are, and being able to support them regardless of location, is incredibly important. Because we're fully GDPR-compliant and have invested heavily in privacy and security, we can help communities operate confidently across borders.

Another key point is how we've designed the platform itself. We made an intentional architectural decision to collect data across disorders, not disease by disease. This is critical because so many rare conditions are being identified for the very first time. These communities don't have an established body of knowledge or a clear understanding of the full spectrum of symptoms affecting patients. By taking a symptom-driven rather than disease-driven approach, we're able to gather foundational data that works for everyone. Then, based on what we see in the community, we can drill down into more specific areas. It creates a much stronger and more flexible framework, especially for newly identified or ultra-rare conditions that are just beginning their research journey.

As a patient advocacy organization ourselves, we work closely with other patient groups who know their communities better than anyone. They bring the trust, the relationships, and the insight into what matters most to families. We bring the tools, infrastructure, and expertise to collect strong, consistent, research-ready data. Together, that creates both better data and more engaged, activated communities. The two factors that are absolutely essential for progress in rare disease.

How does the Global Gene's Corporate Alliance help align the interests of industry, academia, and patient groups, and why is multistakeholder collaboration so key in advancing rare disease outcomes?

Our Corporate Alliance has been part of Global Genes for many years. It brings together biopharma companies, CROs, and other industry partners who genuinely care about rare disease and understand that patient engagement is essential to building a strong ecosystem. This is key

because no single group can drive progress on its own. Neither patients, biopharma, academic researchers, clinicians, nor regulators can move the field forward without each other. That's why these multistakeholder collaborations are so important.

Our current Corporate Alliance workgroup is centering collaborative work around clinical trials. We often talk about patient engagement as "the right thing to do." It absolutely is, but there are also very real operational and financial benefits. Clinical trials are the most expensive part of drug development. Every protocol amendment costs time and hundreds of thousands of dollars, and slow recruitment stretches timelines. All of this affects the runway companies have to get therapies across the finish line. Therefore, involving patients earlier helps avoid those pitfalls.

This collaborative work has involved both our Corporate Alliance and our Global Advocacy Alliance, which includes more than 800 patient advocacy groups. Together, we developed a white paper called "Early and Often", which focused on how industry can involve patients much earlier in the therapy development process. This year, we've taken the white paper a step further and built a practical implementation framework, which is now heading into publication. The point is to move from theory to action and show not just why early engagement matters, but how to actually do it.

For me, even though I spent years working in industry on the diagnostics and data side, I ultimately see everything through the lens of a patient advocate because of my daughter's experience. From that perspective, it's very clear that building strong, connected ecosystems is what truly accelerates progress. When patients, researchers, clinicians, and companies are working in sync, everything moves faster and the chances of getting effective therapies to people who need them go way up.

While building a large-scale research platform, Global Genes has also continued expanding patient and family support services. Why is this dual focus so important?

We have talked about the impact of patient-led research, I want to emphasize just how essential support services still are for rare disease patients and families. The reality is that this journey is incredibly hard. I still remember how overwhelming those early days with my daughter where. Getting the diagnosis, trying to figure out what it meant, and not having a clear path forward. That's why our patient and family support work remains such a core part of what we do.

Our Rare Concierge service is essentially an open help desk where anyone can reach out for guidance. Whether they need help navigating care, finding resources, or simply figuring out their

next step, we continue to offer much needs resources. Having that kind of foundational support makes a huge difference. It helps people get connected to the right specialists and services, and it gives them a place to turn when they're feeling lost.

And then, for those who want to go further and who feel ready to become community leaders or drive research in their disease area, we can help them down that path too. But it all starts with that initial support.

How else is Global Genes building trust and designing programs to better reach and serve these communities?

A big part of it is education. Part of our mission is to create spaces where patients, advocates, researchers, and industry can learn together and connect. For example, we host several events throughout the year, including our Rare Drug Development Symposium, which we held in Boston this year in collaboration with Boston Children's. The goal is to walk advocates through the drug development process in a way that's accessible and practical.

We also run more in-depth programs like our Rare Advocate Development (RAD) Workshops. These are three-day deep dives that really get into what it takes to advance therapy development. Our current program, the RAD Brain Workshop, is focused on CNS disorders and is being run with the Rare Epilepsy Network and Mahzi Therapeutics. Programs like these not only provide foundational scientific understanding, but also help advocates build the networks they need to connect researchers, industry partners, and potential advisors.

Many advocates don't come from scientific backgrounds, yet they're suddenly thrust into roles where they're expected to drive progress for their communities. I often think of them the way I used to think of product managers when I worked in software. They're the ones coordinating across teams, setting priorities, and keeping everything moving. Our job is to give them the tools, education, and connections to step into that role confidently and effectively.

How do you envision the next phase of rare disease research infrastructure and community empowerment and what most excites you about the future?

I really feel like the entire rare disease ecosystem is accelerating. We're still in the early days, but the rise of potentially disease-modifying therapies, and the ability to think about them as platforms

rather than one-off solutions, is going to fundamentally change how we approach research and drug development. Instead of treating each rare disease as its own isolated issue, we can start thinking collectively, just as we've long done with small molecules that treat classes of conditions. The difference now is that these newer technologies may actually modify or correct the underlying biology. That opens up so many new possibilities and I's excited about is seeing groups of disorders begin working together in very intentional ways.

For example, my daughter's disorder is a synaptopathy that affects communication between neurons. I was recently at a synapse symposium in San Diego where researchers were looking across many different genes involved in synaptic transmission. What became clear is that the science is already moving in a more holistic direction, and now several different patient communities are starting to talk about creating their own consortium to match. When groups pool their knowledge, resources, and momentum, progress can move so much faster. That kind of collaboration is going to be an important part of what comes next.

Simultaneously, the other major development that's both exciting and challenging is the emergence of individualized medicines. We're shifting from one therapy for the whole community toward situations where a therapy might be developed for a single individual. These "N-of-1" approaches can be transformative, but they also raise new questions. How will we regulate them? How do we ensure safety? How do these individualized programs eventually roll up into broader therapeutic pathways? The New England Journal of Medicine's article on the plausible mechanism framework gives a hint of how this could work, but there's still a lot to figure out.[1]

What's most important to me is that we don't lose sight of the bigger picture. We need solutions that work for ultra-rare communities with only a handful of patients and for the larger—but still rare—populations that also desperately need options. If we get this right, the combination of scientific innovation, platform thinking, and thoughtful regulatory evolution could open up both real economic opportunity for biotech and biopharma, and more importantly, real therapeutic progress for patients.

What message would you like to leave with policymakers, industry partners, and rare disease communities?

I think the final message I'd want to leave is that keeping patients truly at the center of this work is absolutely essential. Patients need a real seat at the table, and their experiences, urgency, and insights are what push this entire field forward.

If we involve them meaningfully, not just as participants but as partners, it will accelerate everything we're all trying to accomplish. Whether you're in biopharma, policy, research, or advocacy, we all have a responsibility to keep this at the forefront of our work. If we can accomplish this, the progress we make will be faster, smarter, and more aligned with what actually matters to the people living with rare disease.

Sources

[1] https://www.nejm.org/doi/full/10.1056/NEJMsb2512695

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