Turning Words Into Action

The 2022 Global Genes Equity, Diversity, and Inclusion Report

FEBRUARY 2022
A Letter from Our CEO

One of the key messages from the inaugural RARE Health Equity Summit Global Genes held November 18 to 19, 2021 in Philadelphia, was the critical role partnerships will play in addressing health disparities and ensuring improvement to equity, diversity, and inclusion.

The summit itself is a reflection of the role collaboration can play in advancing these issues. The event is part of a multi-year partnership between Global Genes and Rare Disease Diversity Coalition in an effort to better understand persistent gaps in diagnosis, and access to research and care for minoritized and underrepresented patient communities.

We know that people with rare diseases are inherently underserved. Despite the fact that roughly 10 percent of the global population has a rare disease, people with these conditions must endure extended diagnostic odysseys, lack available treatments, and struggle to get their conditions recognized and prioritized. For communities of color and other minoritized populations, the challenges and disparities are even greater for getting a diagnosis, finding and connecting with others in their disease community, being included in clinical trials, and receiving equitable and timely access to treatments and care from medical specialists.

The summit brought together stakeholders from across the rare disease continuum and highlighted many of the issues that need to be addressed in wide-ranging discussions. While these serve as a good starting point, it’s important that we seize this opportunity to now put action behind our words.

Our goal is to eliminate the burdens of rare diseases for families and patients globally. We do this by equipping, empowering and connecting individual patients and caregivers and emerging and established patient communities to take the next step forward in their journey to drive progress within and across rare diseases.

We also share a belief that progress will be based on connectedness between and among minoritized communities and rare disease organizations and other health system stakeholders. We must work together to collect and share information and data needed to identify and overcome disparities in access to diagnosis, trials, and treatments, and ultimately to achieve health equity.

In the months and years ahead, I hope you will join us to not only continue the conversation, but to take action to help address health disparities, and ensure all patients with rare diseases can gain access to the care and treatments that all should have.

D. Craig Martin, CEO
Global Genes
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If we are to reduce health disparities, Monica Webb Hooper said, “we need to bake equity in as an ingredient in all of our scientific and health-care efforts.”

Hooper, deputy director of the National Institutes of Health’s National Institute on Minority Health and Health Disparities, delivered the keynote address at the 2021 RARE Health Equity Summit.

“Advancing the science of health equity means that we are intentional about how we build our scientific programs, our training programs, how we engage with communities, and how we develop and execute the work. It means that we've planned at the outset how we'll make equity as that primary ingredient, so that we can advance this aspiration of health equity, which is not an outcome,” she said. “Equity is the assurance. It is an assurance of the highest level of health for all people. And this requires that we close the gaps, which are disparities—that we value everyone equally, that we address avoidable inequalities, and that we provide supports that are proportional to the needs.”

Hooper distinguished between health differences among different populations and health disparities. While a specific population may have health differences because of inherent biologic or genetic conditions, health disparities relate to issues where the causative agent is disadvantage, whether as a result of social, economic, or environmental drivers. In these instances, the underlying difference in health and health outcomes is driven by injustice.

Health disparities have been documented to show such things as shorter life expectancy, higher rates of cardiovascular disease, cancer, diabetes, infant mortality, stroke, and cognitive impairment. Health disparities have also been tied to differences in prevalence and outcomes in mental illness. “There are considerable racial and ethnic disparities in most conditions,” Hooper said.

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— MONICA WEBB HOPPER, NATIONAL INSTITUTE ON MINORITY HEALTH AND HEALTH DISPARITIES

Disparities in rare diseases include differences in hospitalizations, time to diagnosis, mortality, and pain. In the case of some diseases, such as the rare liver disease primary biliary cholangitis, there is a lower hospitalization rate among African American patients and a higher risk of all-cause mortality. She notes that the lower hospitalization rate could be indirect evidence of lower access to healthcare, suboptimal out-patient management (because this is a condition that develops slowly over time), and it could reflect decreased access to evidence-based care.
Hooper said that many scientists and doctors presume that genetics or biology are responsible for racial and ethnic disparities. Indeed, said Hooper, racial and ethnic minoritized groups have a disproportionate burden of underlying comorbidities.

“The problem with these explanations is that they are absent the full context of health and wellness and illness and disease,” she said. “And they’re absent the recognition that race is a socio-cultural political construct, not a biological or genetic one. And race is important to study because it shapes the lived experiences of all groups, but it’s important to study at a social level.”

It is that full context, which includes systemic factors, such as historical and ongoing discrimination, and the effect of chronic stress on health, which she argues is important to consider. This is also true within rare disease. She said researchers need to consider the effects of what happens on the outside of the body to what happens inside of it.

One concern is that researchers may conflate racial differences—differences in genetics and biology—with health disparities.

“Many people have raised this concern that this perpetuates a false belief by some that differences in disease outcomes stems primarily from inherent pathophysiological differences between what really are social categories,” she said. “Researchers and community leaders have raised concerns that the consequences of the reliance on the biology of race are many. And they lead to the promotion of things like eugenics, misattributions about health and disparities, and the lack of progress in addressing the root causes of disparities.”

As a reflection of the ability to address health disparities, Hooper pointed to the NIH Community Engagement Alliance Against COVID-19 Disparities, which was established to lead outreach and engagement efforts in racial and ethnic minoritized communities that have been affected disproportionately by COVID. While the lack of diversity in clinical trials is an ongoing concern, Hooper pointed to the work NIH did to help Moderna enlist a diverse group of clinical trial participants for its vaccine study. About 20 percent of participants in the study were Latinx and 10 percent were Black.

To achieve that, NIH offered direct guidance about evidence-based strategies to increase enrollment of underrepresented populations, such as extending the clinic hours, having clinics on the weekend, setting specific targets and swift follow-up with individuals who expressed interest in participating.

With regards to the study of rare disease, Hooper said it’s important that we ensure diversity of our studies not because we expect genetic differences by race or ethnicity, but because everyone should have the opportunity to receive cutting-edge care and participate in the process of generating medical discoveries. She said we need to recognize that people who are racial and ethnic minorities in general are just as willing to participate in clinical trials.

“One suggestion that works and is often forgotten,” she said, “is we just have to ask individuals to participate.”
Hannah Barnhart spent a number of years in Alaska, most recently working as a clinical coordinator for Providence Health & Services in Anchorage. She recalls the barriers to healthcare the native population there faced as a result of geography and the distance people would need to travel to get care.

“For some of the patients I’ve dealt with, we actually had to arrange their transportation to the small float plane, from the float plane to the big plane, from the big plane to the taxi, from the taxi to the hospital, and then from the hospital to the lodging,” she said. Some of the patients have never been to a big city before. This is their first time.”

In addition to the “newness” of things like MRI machines that patients may have encountered for the first time, there were cultural barriers. It’s common for an Alaska native patient to ask a health worker where they are from. Though health workers may find such questions off-putting, she said it’s a way for them to get to know you and include you in their circle of trust.

Barnhart’s comments came in a panel discussions about reducing health system biases faced by rare disease patients in underserved populations. Pediatric Resident Physician at UNC Health Care Christel Wekon-Kemani, Associate Center Director of Patient-Centered Initiatives and Health Equity at the GW Cancer Center Mandi Pratt-Chapman, and Chief of Operations for the Global Partnership for Telehealth Sherrie Williams.

One way to address the barriers underserved populations face is to turn to telehealth. That’s particularly true in a place like Alaska, but the panelists were quick to note that while there have been some successful pilot projects, people who live in remote areas often lack access to broadband and other technologies, and face other barriers.

“If we think about our medical schools, we’ve not prepared our new doctors and nurses and other healthcare professionals coming out with that knowledge, that foundation that we can treat patients virtually,” said the Global Partnership for Telehealth’s Williams. She said her organization is working with medical schools to create curriculums that incorporate telehealth, but that many providers remain hesitant about delivering care in this way. She pointed to surveys and said while patients are overwhelming in favor of the use of telehealth, only about a third of providers said they are willing to offer it.
“I think this is a temporary barrier,” she said. “As we continue to have this conversation around technology and how to reach patients who are unreachable, we’ll solve this problem, but it can be a barrier now.”

GW Cancer Center’s Pratt-Chapman, who recently published a study on the use of telehealth with cancer survivors during the COVID pandemic, found contrary results with providers being more willing to embrace telehealth than patients, but noted they indicated a lack of training in using telehealth and worried about missing something during interactions with patients.

Kemani, who works as a physician in rural North Carolina, said he feels bias play a major role in the diagnosis and management of rare diseases, especially in marginalized patient populations. As an example, he said he’s seen socioeconomic biases influence the type of options a provider will present to a family. He said a provider may make assumptions about what a family can afford based on their insurance. This may lead a provider to offer hemodialysis rather than peritoneal dialysis because they assume a family can’t afford the more expensive option.

He also believes there is a problem with implicit bias providers have towards patients from certain populations, particularly in interactions with minoritized populations. He said these preconceived notions can be hurtful and harmful and can lead to poor health outcomes. As an example, he pointed to what he’s seen and heard from patients and families of children with the rare blood disorder sickle cell disease.

“These patients—more so on the adult side than on the pediatric side—are labeled as drug seeking, labeled as difficult. These patients are called names, such as sicklers,” he said. “That term creates the idea that their disease defines them as people. In fact, these people just have this chronic disease that they’re living with and there’s so much more to them than a disease.”

He said in such instances, care staff may avoid going into rooms with these patients because they are viewed as difficult or drug seeking. That in turn lowers the quality of care they receive and contributes to poorer outcomes.

Last year, Kemani worked with others to implement a medical education curriculum for residents to address bias through a four-part conference series that addressed medical bias, racism, and health equity. “Everyone, whether they believe it or not, has implicit bias,” he said. “[We need to] make sure that they’re able to identify that, and to realize that, and be aware of that as they interact with patients of all backgrounds.”

GW Cancer Center’s Pratt-Chapman said she conducts implicit bias training. “I always tell people that everyone has implicit bias, and we make mistakes if we’re trying to get outside of our comfort,” she said. “When I give trainings, I try to be very clear. I’m a cisgender woman that doesn’t represent all LGBTQ people. I can tell you what I know from my research, and I can tell you from my personal lived experiences, but we’re not monolithic.”

The panel’s moderator, Price, noted that none of the things the panel spoke about happen in a silo.

“None of these things are happening independently of each other, but they’re all at play—interconnected,” she said. “When we think about what actions we’re going to take, what conversations that we need to be having, we’ve got to make sure that it’s this multi-pronged kind of approach and viewpoint to look at it.”

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— CHRISTEL WEKON-KEMANI, UNC HEALTH CARE
Grace Whiting was talking about the problem of unconscious bias when she pointed to an article from the investigative news nonprofit ProPublica that came out in the midst of the pandemic that included an anecdote about an African American man who went to an emergency room, but doctors ignored his and his caregiver’s complaints. He was sent home to die.

She said the article illustrated the great disparity in care because of the way that medical professionals might react to someone based on how they look.

“Women will report time and time again they’re in pain, or that they’re experiencing symptoms, and those will get minimized. That creates a lot of barriers, especially when it comes to rare disease, to have someone come into the office and say, “No, I know my child. I know my self. I know myself. I know my partner, and I know that this is not normal. And I need you to trust my experience so that we can find a solution,”” said Whiting, CEO of the National Alliance for Caregiving.

“A lot of times, patients and their family members experience that type of minimization of concerns where you’re having to argue with your physician about whether or not what you’re experiencing is real.”

Whiting made her comments during a panel discussion on the diagnostic odyssey for rare disease patients in underserved communities during the 2021 RARE Health Equity Summit. National Center for Advancing Translational Sciences Program Officer Eric Sid, and Angioma Alliance Health Equity Program Development and Outreach Specialist Jessica Biggs joined Whiting for the session, which Associate Director of Patient Advocacy for Travere Therapeutics Sika Dunyoh moderated.

Among the topics the panelists discussed were the barriers members of underserved communities face in getting a diagnosis and what stakeholders could do to address them. Whiting suggested to start with what people know works well, by finding clinicians, researchers, and practitioners who have engaged these populations to learn from them. She pointed to research done by Children’s National Hospital on engagement and outreach to diverse populations in the Washington, D.C. area. They have people who are designated to sit down, talk with
people about what does it mean if you engage in this research? She said such a person-centric approach is important across the whole healthcare system because “it’s not a one-size-fits-all solution.”

NCATS’ Sid recalled discussions about “cultural competency” during his training and seeing it described in a textbook. He has come to dismiss what he called “easy solutions” because, he said, it’s not possible to become competent in a culture by attending a lecture or reading a book. Rather, he said, what is needed is “cultural humility” and “understanding what we can understand, and what we don’t understand, and just being open to things.”

He said in his own programs, with regards to providing public health information in the past, the focus had been on the use of plain language, information that anybody can read at an eighth-grade level and understand. Now, though, he said the focus instead is on health literacy.

“We’re thinking, not just can you read this information? What can you do with it? Who are we targeting? Who has access barriers perhaps with language, perhaps even with the verbs that we’re using, the choice of pronouns?” he said. “What we need to be thinking about really is how we make that information applicable to not just everybody, but to those that have the most need of these resources in particular.”

Angioma Alliance’s Biggs said she spends time with different communities talking to them about their needs and the stressors in their lives and she said the important thing is always to be listening. “Trying to learn as much about your patient,” she said, “listening to your patient, and building that trust that way.”

One final point the panelists made was that it is action, not gestures, that are needed. Whiting recounted a recent conversation she had with an organization that asked her about what they could do to improve diversity and inclusion. Whiting noted that the organization’s budget line for diversity and inclusion was $0.

“You could start investing in these programs,” she said. “That’s the first step. Instead of saying that’s something we can cut and we don’t need to have a representative research sample. You start from the position of, this is a requirement, and we know it’s going to cost more money and we’re willing to invest.”

Sid echoed that point, saying that we need to consider all the actions we have across the entire system, including who’s involved in the conversations, and make sure we have more rare disease providers and researchers who are from the same populations that they’re serving. He said this would ensure that when you’re thinking about the study protocols that are being developed, or you’re thinking about the problems in terms of the data collection, are you thinking about the issues in terms of building that rapport and trust, “that we have people there that understand what those issues are in the community and are able to both bring that into their own job and role, but also into their scientific field or their practice as well.”
Researchers, who were studying people who were deaf and suffered from schizophrenia in an effort to understand the neural pathways involved when people hear voices, found one deaf patient who heard screaming, which lead to a number of papers that implicated a part of the brain that processes language in the condition.

Alexa Berk King, head of research and real-world evidence for the consumer health technology company Ciitizen, said it was an interesting finding until a researcher who was deaf explained that the scientists had misinterpreted what the deaf patient was reporting. Rather than hearing screaming in the auditory sense, the patient meant “signing furiously.”

“There’s lots of opportunities to build trust in a community, not just in the medical setting, but beyond as well.”

— CLARE THIBODEAUX, CURES WITHIN REACH

Her comments came during a session on diversity in rare research during the 2021 RARE Health Equity Summit. Vice President of Scientific Affairs for Cures Within Reach Clare Thibodeaux, and Life Sciences Lead for Amazon Web Services’ Open Data Team Erin Chu joined King. Isaac Rodriguez Chavez, senior vice president for scientific and clinical affairs for the clinical research organization ICON, moderated the panel.
One of the themes that came up during the discussion was the need for researchers to build trust in underserved communities. Cures Within Reach’s Thibodeaux said when building trust, it is critical to engage with community organizations and think beyond just those in healthcare, whether it is religious organizations, social services organizations, or leaders in a community.

As an example, she pointed to a speaker at a recent Cures Within Reach event who was designing a study that focused on lung cancer within the Black community. In assembling a multi-disciplinary team, he not only turned to principal investigators from different health centers, but also included a local barber.

“If this barber became what he called a citizen scientist,” she said. “This was somebody who had daily contact with the community and was able to talk to them about clinical research in general, tell them about this trial, and encourage them to learn more. There’s lots of opportunities to build trust in a community, not just in the medical setting, but beyond as well.”

When Rodriguez asked the panelists why it was important to include equity, diversity, and inclusion in research, Amazon Web Services’ Chu spoke in personal terms. She said her father was diagnosed with Moyamoya disease, a rare cerebrovascular disorder caused by the blockage of arteries at the base of the brain. The condition is known to be more prevalent in Asian populations.

“If you don’t have equitable access to data or equitable representation in data, then your baseline is skewed at the very beginning,” Chu said. “What I’ve been dedicated to is increasing not only the ability for people to access data that are in the public domain or in some cases in a controlled but still distributed area, but also enabling the data provider to be empowered to share the data and give people access to build on that data.”
In discussing the social determinants of health, Shonta Chambers, said it is time for us not just “to be transactional, but transformative.”

Chambers, executive vice president of health equity initiatives and community engagement for the Patient Advocate Foundation, a nonprofit that provides case management services and financial aid to Americans with chronic, life threatening, and debilitating illness, said that we live in a society where the political structure continues to perpetuate systemic racism that has fortified disparities in the quality and affordability of healthcare.

“They’re under this umbrella that we call the social determinants of health, and they include everything from looking at economic stability, to housing, to neighborhoods, everything that one needs to be healthy,” she said. “When we talk about these social determinants of health, we have to come down a little further and understand the social risks that actually have been caused by unaddressed social determinants of health. When you don't invest in a neighborhood, then how can you expect someone to have transportation, have the infrastructure that they need, have the broadband infrastructure that they need, have the health system that they actually need?”

Chambers comments came during a 2021 Rare Health Equity Summit panel discussion on expanding access to underserved patient communities. Program Director of the O’Neil Comprehensive Cancer Center at the University of Alabama at Birmingham Claudia Hardy and Managing Member of The Watts Group Daron Watts joined Chambers for the panel. Pfizer Biopharmaceuticals Group Regional President of North America for Rare Disease Bert Bruce, who moderated the panel, framed the discussion around the question of what changes need to be made so that underserved patient communities can gain greater access to care without requiring them to do anything differently than they are doing today.

Hardy said while it’s good to have discussions about social determinants of health and issues like health equity, discussions in the abstract will have little meaning to people living with the reality that they have to catch a bus to see their doctor, wait for a child to get home from school before going to an appointment, or need to rearrange their work schedule or lose pay in order to get care.

“Social determinants of health are powerful, but I caution all of us who are the practitioners to take a step and understand the patients, the individuals
that you’re working with, and that you’re trying to serve,” she said, noting many patients don’t know to ask whether their insurance covers a drug they are prescribed, or to ask for a social worker to be assigned. “There are so many different layers, but many of us are not speaking the same language. All of these are things for consideration, both for the user and for the practitioner. We have to think this thing through and ask the powerful questions so that we can get the total picture of what we have represented before us today.”

Watts, who is a regulatory attorney, focused on the need for easing the regulatory pathway for medicines that treat rare diseases and the access issues created by organizations like Institute for Clinical and Economic Review (ICER), a nonprofit that issues reports on the value of medicines and is widely used by payers to determine what medicines they will cover and at what price. He said ICER in the past has erected barriers to access by applying a health technology assessment system that is ill suited for rare disease that disproportionately affect people of color.

“They are truly those gatekeepers, those frontline individuals that are often the first people that get a call as an individual, as one is trying to access a health care system,” said UAB’s Hardy. “What it really comes down to is how do we really fortify the work that they do by putting in their toolkit additional resources to help them as they’re working with patients and link them to local and national resources.”

She noted that the O’Neil Comprehensive Cancer Center began with volunteers and then with federal funding they were able to hire and integrate these community health workers so that once a patient is diagnosed, they are assigned a community-based navigator to help guide them within the system.

The Patient Advocate Foundation’s Chambers said as COVID has played out, it’s begun many conversations about health disparities. “Let’s stop having the conversation,” she said, “and let’s get to some solutions.”

DARON WATTS, THE WATTS GROUP

CLAUDIA HARDY, O’NEIL COMPREHENSIVE CANCER CENTER

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Learning Cultural Humility

To reduce healthcare disparities in the healthcare setting, Ricardo Correa said it is critical for healthcare providers to understand people from other cultures.

Correa, program director of the endocrinology, diabetes, and metabolism fellowship at the University of Arizona College of Medicine, Phoenix, made the remarks during a keynote address on the second day of the 2021 RARE Health Equity Summit where he spoke about the importance of cultural consideration in healthcare.

“The first thing is cross-cultural education. That’s the key to everything,” he said. “It is understanding how the other culture works, how the other culture has different things, and trying to provide care in that kind of culture.”

He said there are some models that are in place that have been working well. The cultural competency model takes the approach that when healthcare providers understand what a different culture believes and what their values are, providers can address a patient that has a different background.

Correa, who spends a lot of time serving the Latinx community, said there is a lot of belief that a superior force will cure them, and they often start praying. Priests, as well as doctors, are an important source of information in the Latinx culture, he said.

“‘It’s important to understand that and not assume that this patient doesn’t want to take care of themselves,’” he said. “‘Because cross-cultural efficacy is when you understand your background, and then what bias you can have, and then communicate with a person of a different background. And finally, the cultural humility is when you do self-reflection and you try to improve.’”

He said every time a doctor comes to see a patient, it’s important that they ask themselves, who is this patient that I’m seeing? What is their background? What is the patient population that patient belongs to? Doctors need to accept some social behaviors of that patient independent of where they are coming from and respect their cultural health beliefs. Oftentimes, he said, there’s not a lot of cultural sensitivity in the healthcare system.

Correa noted that from group to group, there are different perceptions of what health and illness are. For instance, he volunteers in a clinic that treats a
population that includes many people who are undocumented and have a belief that the grandmother is the one that has the solution for everything. "We have to understand that and start working with the grandmas, empowering the grandmas on medicine," he said.

He contrasted the societal determinants of health—issues like systemic discrimination—as being different than the social determinants of health. He believes that a healthcare system has a responsibility to have linguistic and cultural competency to serve its patients. That, he said, requires seeing patients as more than their medical problems, but as human beings in the context of all the things that can be happening around them. Bridging language barriers with a translator is not enough to bridge the cultural issues. Using a translator doesn’t solve the cultural problem. If doctors don’t understand their patients’ culture and don’t understand their unconscious bias from their own culture, nothing will solve the issue.

Many studies have shown that providers who are culturally competent provide better care to their patients. When there is cultural insensitivity the quality of care will decrease. Correa said this is important because of the demographic changes the United States is undergoing with the Latinx community growing to be the largest minority population in the United States and Caucasians becoming a minority as African American and Asian-American populations grow as well.

It’s not just about race. Correa has been involved in establishing a center of excellence for transgender patients with endocrine issues. There were some healthcare professionals that decided because of their own biases, not to see patients in that clinic, in the belief that care would suffer as a result. The clinic set up a coaching system with 10 people to a coach, who guides them on working with transgender patients.

“We have focused on training the ones that want to get trained,” he said. “We have enough people that want to get trained. And with that, we can provide better care for them.”
He said Pfizer was “intentional and specific” in looking to investigator sites in diverse communities that were disproportionately affected by the pandemic. But once enlisting a site, it impressed upon investigators the importance of recruiting individuals who mirrored the racial and ethnic diversity of their communities. The company also raised awareness with advocacy organizations and integrated targeted public relations that looked to encourage minority participation in the COVID vaccine trials.

“We didn’t do any of this alone. Community education, community outreach—how we partner with communities to build trust (or a phrase that I sometimes use, which is borrowed trust), because organizations exist in these communities that are committed to these communities well before we arrive,” said Bruce. “That aspect of partnering with them and having them trust in who we are and what we’re looking to do, and then commend that trust to the communities they partner with, we believe is one of the shorter ways to be able to work effectively together.”

Pfizer assembled what it called a multi-cultural health equity collective that worked with several organizations. As a result, of the approximately 46,000 individuals who participated in the initial phase 3 vaccine clinical trial, about 42 percent of those were global and more than 30 percent had racially
and ethnically diverse backgrounds. He said sustained partnerships with more than 35 advocacy organizations helped the company achieve its health equity and diversity goals for the COVID-19 vaccine trial.

“All of these efforts really don’t mean very much unless people have actual access to the vaccine regardless of where they live or what their circumstances are,” Bruce said. He noted that the company is working with global stakeholders to address supply chain issues in low-income countries to understand how the private sector can lend expertise and support in delivering COVID-19 vaccines. The company is working with the World Health Organization and has pledged to provide a total of 2 billion doses of its COVID-19 vaccine to low- and middle-income countries in 2020 and 2021.

Bruce said the pandemic highlighted the inequalities that exist in our healthcare system and society, and has sparked a number of new conversations.

“When I reflect on our experience with the pandemic, one of the key takeaways is that equity has to be a focus throughout the entire process of advancing new medical innovations from the early discovery stages, through the development stage to delivery and ensuring access,” he said.

Bruce likened the inequities made visible in the pandemic to the inequity that is characteristic of rare diseases as patients face unique challenges and barriers to care, from delays in obtaining a diagnosis, and often, little or no treatment options available.

“If we don’t address these inequities across the spectrum from diagnosis to access, then patients simply won’t benefit from treatment innovations,” he said.

Bruce pointed to a number of instances where Pfizer is applying the same type of outreach it did with its COVID vaccine to reach underserved patients with specific rare diseases including transthyretin amyloid cardiomyopathy, which is often underdiagnosed in the Black and Afro-Caribbean communities and can cause heart failure. The company is doing outreach, education, and partnering to raise awareness about the condition.

In a separate example, he said through discussion and engagement with advocacy groups, healthcare providers, and patients, Pfizer learned it needed to better reach women living with hemophilia who face distinct challenges, particularly when it comes to diagnosis. The company partnered with both national and grassroots advocacy organizations to support their efforts to better reach this underserved community, including the launch of a new website, offering education and resources specifically for women impacted by hemophilia.

And, in order to raise awareness and dispel myths with regards to sickle cell disease, the company engaged in several grassroots partnerships, including one with the National Newspaper Publishers Association, a trade association of more than 200 African American-owned community newspapers in the United States and Virgin Islands.

“COVID-19 has taught us that meeting these challenges head on is possible, and that science will win,” he said, “Pfizer is committed to working to address the inequalities in the systems for rare disease patients through collaboration, with patient community transparency, diversity in our clinical trials, and a renewed focus on equitable access.”

Equity has to be a focus throughout the entire process of advancing new medical innovations from early discovery stages, through development stage to delivery and ensuring access.

— BERT BRUCE, NORTH AMERICA FOR RARE DISEASE, PFIZER BIOPHARMACEUTICAL GROUP
The demographic make-up of the population is shifting and with that so too is the burden of disease in the United States. Already five states have minority-majority populations and by 2045, the nation will be a minority majority country.

Joy Russell, vice president of external affairs for Genentech, in a presentation at the 2021 RARE Health Equity Summit noted that by 2030, new cancer cases will increase 80 percent in low-income countries. In the United States, by 2045 more than half of the patients living with Alzheimer’s disease will be Black or Hispanic. And by 2050, the number of cases of Alzheimer’s disease in China is expected to quadruple and account for half of all cases worldwide.

“It really underscores the work that we have left to do with patients to have equitable access and experiences in our healthcare,” said Russell, who noted a recent report Genentech had done that found 54 percent of medically disenfranchised patients feel the healthcare system is rigged against them. “I know that makes us all very sad. It certainly does from an industry perspective as well. We know that we are absolutely seminal in that change. We have to really bring the best interests of society as we look at policy development for equity, as well as how does that really meet the patient where they are in their journey from rare disease.”

Russell said that to bring about change it’s going to take industry, advocates, and the community at large. As part of Genentech’s commitment to health equity, she said it is working to build and strengthen partnerships across its communities. The company is also looking for ways to embed diversity and inclusion in its work. As part of that effort, that means building a diverse and inclusive team of employees.

“We want to advance inclusive research and health equity,” said Russell, who noted that just 2 percent of patients in Alzheimer’s clinical trials are diverse. “That’s a problem. We have to start fixing and looking and asking deeper questions as an industry. And through that, we will transform society.”

In breast cancer, the numbers are also telling. Black women have a 40 percent higher mortality rate than white women, but when it comes to clinical trials, represent just 6 percent of study participants.

Russell said the company is engaged on a number of fronts to improve health equity. For the first time Genentech has mandated that officers across the company put together action plans around how we embed diversity and inclusion and foster a sense of belonging as an employer.

The Genentech Foundation has invested $90 million, 48 percent of which has gone to support underserved communities. It directed more than $16 million to tackling systemic barriers and $6 million to broaden access to essential health care related services.

And, with an election year in 2022, the company is looking to the policy front to develop better access to care through legislation and patient advocacy and is working with patient groups and health equity advocates to do so.

“The treatment and the access that we know that we have to provide,” she said, “is very incumbent on partnerships and partnering with folks all like you.”
Though many people in the rare disease community are all too aware of the health disparities that exist throughout healthcare prior to the COVID-19 pandemic, it nonetheless served as a “much-needed reminder of the disparities in our healthcare system,” said Eve Dryer.

Dryer, vice president of patient advocacy for Travere Therapeutics, said in the rare disease community, patients of color often face longer wait times to see a doctor, are less often referred to specialists, and are far less likely to be provided information to participate in early research or clinical trials for potential new therapies.

Speaking at the 2021 RARE Health Equity Summit, she noted that people discussed plans for cultural sensitivity training for providers to help address some of these issues for clinical trials, tools for education to better engage patients of color, and potential policies to accelerate rare disease diagnosis and treatment in diverse populations. She said the question is “How do we work together to turn these ideas, these possibilities, into reality and into action?”

“What we are talking about today is not about applying superglue to the cracks in the foundation. Rather, this is going to have to be about rebuilding the foundation entirely,” she said. “Why? Because, as the esteemed Nobel Peace Prize winner Desmond Tutu once said, ‘There comes a point where we need to stop just pulling people out of the river. We need to go upstream and find out and address why they’re falling in.’”

She said health inequities in rare disease are at the forefront of discussions that are taking place in Congress, in the medical and science community, in patient organizations, and in industry. She said it’s imperative that any actions lead to real impact and that everyone work together to find tangible solutions and not just gradually chip away at obstacles.

To achieve health equity in rare disease requires first understanding the unique challenges people living with rare disease face, especially diverse patients, and then starting to address them.

She said her team at Travere works hard to integrate the perspectives and experiences of diverse patients and caregivers into the drug development process through numerous channels. This includes its patient...
and caregiver advisory council, quarterly community conversations with advocacy leaders, and its diversity advisory boards. It also actively engages in supporting diverse representation in its clinical trials.

The company has also underwritten and launched diversity health initiatives, such as the 2021 RARE Health Equity Summit and other rare disease and diversity health organizations including Everylife Foundation’s diversity fellowships and Uplifting Athletes’ efforts to help launch the underrepresented researcher awards, and as the founding sponsor of the Rare Disease Diversity Coalition. The company is also working with organizations focused on specific rare diseases to build their health equity initiatives.

The company issued a total of $500,000 in grants to address delays in diagnosis, improve engagement between healthcare providers and patients, and understand what must happen to make sure that research reflects that diverse populations and participants of color are equally represented in those efforts to launch clinical trials.

To turn the ideas into action, though, Dryer said will require everyone getting involved and catalyzing others to recognize the need to do the same. She encouraged people to join the Rare Disease Diversity Coalition or support other organizations that are working towards the same goals of advancing equity and diversity in rare disease.

“As we head down this path together and roll up our sleeves to really drive this work forward, I ask that we hold ourselves and each other accountable,” Dryer said. “I ask and urge you to lean on each other for help because the road ahead of us in achieving equity will not be easy, but it can and it will be done with the combined knowledge and passion of those of you here today.”
During the 2021 RARE Health Equity Summit attendees participated in four simultaneous workshops that covered improving rare health literacy, challenges adults and older rare patients face, challenges for young adult rare patients, and addressing racial and ethnic disparities. Following the workshops, Rodney Samaco, assistant professor of molecular and human genetics at Baylor College of Medicine, moderated a panel discussion with workshop leaders to share with the full summit attendees highlights and takeaways from these sessions.

On health literacy, Samaco said in the past, there was a focus on the individual and their ability to access, understand, and use information to make good health choices and access services. Now, he said, a new dimension has been added to health literacy to take some of the onus away from individuals and consider organizational health literacy, the degree to which organizations and institutions are equitably enabling individual health literacy.

The groups that met on this topic devised overlapping solutions. One group focused on how to make educational materials for rare patient populations more accessible and understandable. That group also discussed ways to make materials clear, but culturally competent, culturally relevant, and culturally sensitive.

Its solution was to put into place an action plan for how to go about redeveloping educational materials. In doing so, he said the group found that it needed to engage with different stakeholders. At the top of that list was the community that they were trying to serve, and also health information experts in that particular disease community including scientists, physicians, and drug developers. Their process included working together with those groups, but then doing extensive user testing of the materials on a wider audience to make sure they achieved what they set out to do.

The other group focused on how to get physicians and healthcare providers to better meet the needs of their rare disease patients. That group’s solution was to work with the ECHO program, which provides free continuing medical education for physicians. It encourages
them to engage and learn about different diseases so that they are educated about them. Their solution was to raise awareness about the availability of this program and get rare disease communities to form coalitions to access and navigate the process of applying for grants in the program that is offered by ECHO.

Serita Edwards, CEO of the E. WE Foundation, who co-moderated the session on addressing rare and ethnic disparities, said much of the group’s discussion focused on the need for change when it comes to rare disease management and optimizing better healthcare outcomes for patients with rare diseases. The solutions the group devised centered on collaborations between patient organizations, community-based organizations, practitioners, and the pharmaceutical industry. The focus of these collaborations would be to raise awareness and educate various stakeholders on the importance of diverse representation on all platforms.

Linda Thomas Wade, CEO and co-founder of Sickle Cell Association of Texas Marc Thomas Foundation, who co-moderated the session with Edwards, added that the group also discussed the need for better communication, lack of resources, and diversity in clinical trials. Among the solutions the group discussed was the need to form meaningful relationships to change the status quo. The group also discussed the need to build trust with the community at the clinical level, which she said is important, individualized patient-centered care, early engagement, and willingness to challenge the status quo.

In the workshop about young adults, one of the key points the group focused on was the transition from pediatrics to young adulthood and finding the right doctor who specializes in a patient’s particular rare disease. Dominique Goodson, president of the SCD Forum, said many times parents and doctors may not have talked to them when they were younger.

She said another key point had to do with language and cultural barriers. Rare disease information is most often in English and there may not be a translation available in Spanish, Arabic, Chinese, or other languages.

Anna Laurent, head of programs and initiatives for Our Odyssey, who co-moderated the session with Goodson, said the action plans were wide ranging. Among those were the call for developing a best practices guide for serving young adults created by young adults for young adults and large organizations. Another action plan focused on the need to make implicit bias training more than a checkbox, as it is for some businesses, and more of a workshop approach.

For the final workshop on adult and older patients, Terri Booker, an attorney and sickle cell patient advocate with Sick Cells, said the group discussed the need to have a rare adult summit tour because once children with rare diseases become adults, nobody seems to care anymore and they are overlooked. She said the group envisioned a tour that moved around the nation and brought together stakeholders and rare disease patients at all stages of their journey.

Nancy Kessler, an independent consultant who co-moderated the workshop with Booker and has an older sibling who was diagnosed at age 64 with a rare genetic condition, said DNA testing is important for the older population. It not only saves lives, but it can be life changing. It can also save the healthcare system money and streamline care. She said we need to better educate stakeholders and create public awareness about the changing medical needs of aging adults. This can be done through everything, from webinars to using the news media.

As the panelists discussed moving beyond words to action, talk once again turned to the importance of collaborations to achieve greater equity, diversity, and inclusion. E. WE Foundation’s Edwards said organizations that are trying to achieve these goals within themselves can turn to organizations that have already been successful and reach out to them to have a conversation.

“You may not just jump right in and say, let’s work together, but it starts with at least having coffee and talking about, is there an opportunity for us to work together?” she said. “It sometimes takes reaching outside to correct something or do something different.”
The way rare diseases are defined vary from country to country, but Lara Bloom, president and CEO of the Ehlers-Danlos Society, cut through the statistical differences and offered a patient’s view. “When I think of rare, I think harder, I think longer, and I think tougher. And why is that?” she said. “It’s because pretty much every healthcare system around the world is set up for the acute, not the chronic; for the common, not the rare. And you see that in economics and care, and it’s just not good enough.”

Bloom spoke on a panel that considered the issues of equity and diversity globally with regards to rare diseases. Founder and CEO of Jeeva Informatics Solutions Harsha Rajasimha, Post-Doctoral Policy Fellow in the DST-Centre for Policy Research at the Indian Institute of Science in Bengaluru’s Mohua Chakraborty Choudhury, and Associate Professor at the Baylor College of Medicine Joseph Lubega joined Bloom on the panel. Executive Director of U.S.-Africa Inclusive Research Strategy for Genentech Markus Gemuend moderated the panel.

Gemuend, who has spent the last five years in Sub-Saharan Africa, said he’s looked at the barriers that patients face, particular patients with non-communicable and rare diseases. In those countries, he said 90 to 95 percent of patients are underserved from multiple aspects. He said that has shaped his belief that health system issues really need to be addressed in a holistic way.

Baylor’s Lubega said, from a global perspective, there are strong geographical differences in the prevalence of rare diseases. What is rare in North America or Europe may be more common in Asia or Africa. “If you’re the minister of health in India, if you don’t recognize that thalassemia is a very common condition in India, but very rare in America, you will end up not paying attention to a disease that’s affecting a huge proportion of your population, which unfortunately is happening.”

As an example, Lubega pointed to sickle cell disease, which is considered rare in Europe and the United States but affects 2 percent of babies born in Africa. Even though it is so common, UNICEF does not list it among the causes of mortality for children in Africa. When he asked a UNICEF official why that was so, the official explained it as a function of funding. “The answer was, ‘Well, I guess the funding we get from the UN, which comes from Europe and America, which considers it a rare disease. So, we don’t focus on it,’” he said. “You would think that UNICEF would know the communities in which they are working and pay attention to conditional sickle cell disease.”
One area of discussion focused on the role of research and clinical trials in making a change in rare diseases in low- and middle-income countries, which the panelists argued would benefit the whole community by giving greater insight into common diseases, as well as providing a better understanding of the diversity of rare diseases.

The DST-Centre for Policy Research’s Choudhury said diversity adds value to any kind of research in any field, but it’s particularly important in clinical research and especially for rare diseases, which by definition affect a small and geographically disperse population. She noted that 60 to 70 percent of clinical trial participants, globally, are white males. And, she said, more than 70 percent of clinical trial research happens in the developed world.

“We can see a significant lack of representation from the developing countries and just not the countries as such, even the communities and people, staying across even in the developed world. So, in terms of science, scientifically, it is also very crucial, to understand how a new drug or therapy will affect patients of varying ages, genders, and ethnic backgrounds, as reactions to many new treatments have proven to be different depending on the individual's demographics,” she said.

When asked what could be done to improve participation in research across the globe, particularly in developing countries, Bloom said it starts with industry being “intentional” with who it enrolls in research studies. She said companies often attend conferences or speak at webinars in the Ehler Danlos syndrome and hypermobility spectrum disorder, even though these conditions are not race- or sex-based, and use PowerPoint slides in which patients are represented by white women. She said companies need to ensure that they are capturing the range of patient stories and that speakers are presenting diverse examples and case studies.

As an example, she pointed to a study her organization has been working on for two years to understand the pathogenic variance related to hypermobile EDS, the most prevalent form of the condition and the only one without a known variant. While the organization began enrolling participants at conferences, it wasn’t a diverse enough population as it only included people who could afford to travel and had the physical ability to travel. In seeking to address the challenges of COVID, the organization turned to lab service companies that would go to participants’ homes all over the world to do blood draws for whole genome sequencing.

“That challenge led us to a much more diverse and inclusive study. And for that I'm grateful for the pandemic forcing us to do that,” she said. “If we continue to keep taking the lessons that we can learn from this, and the challenges, and constantly be pivoting and moving along with the times, then at that point of enrollment that representation will continue to keep diversifying.”

DST-Centre for Policy Research’ Choudhury said there’s a need to conduct more clinical trials in India to increase diversity, but this requires action at the state and federal level to better equip the nation to participate in such studies.

Jeeva’s Rajasimha, who did post-doctoral research at the National Institutes of Health, said he was struck by the lack of diversity in the genetic database. “Medicine is increasingly data driven and patient recruitment included,” he said. “If patients represented in these databases are not diverse, we can’t expect good recruitment because these databases are where we check for inclusion/exclusion criteria—how many patients match the criteria for a given clinical study?”

That’s why he said he’s been working to educate patients about why it’s in their best interest to be in these public databases in a de-identified manner. That, he said, is important in the developed world. In the developing world, electronic health records and medical claims databases don’t exist.

“That type of data access is lacking in low resource environments,” he said. “That poses a big challenge.”

MOHUA CHAKRABORTY CHOUHURY, INDIAN INSTITUTE OF SCIENCE IN BENGALURU

JEEVA RAJASIMHA

MOHUA CHAKRABORTY CHOUHURY, INDIAN INSTITUTE OF SCIENCE IN BENGALURU
Global Genes CEO D. Craig Martin quoted the author Virginia Burden Tower, who wrote “Cooperation is the thorough conviction that nobody can get there unless everybody gets there.”

“Our work is really designed to make sure that everybody gets there,” said Martin. “There are too many being left behind. There are too many being left out, too many being marginalized, and we have an opportunity in a community with a tremendous amount of need to make an impact.”

To do that, he said, it will be necessary to think big, think creatively, and not to stop until the job gets done.

Martin talked about how Global Genes works with individual patients and caregivers who turn to the organization, even some of whom do not yet have a diagnosis. His comments came during the closing session of the 2021 RARE Health Equity Summit. Black Women’s Health Imperative President and CEO Linda Blount, the organization’s Chief Policy Officer and Senior Counsel Tammy Boyd, joined Martin on stage.

“It’s important for us to have partnerships and places that we can help direct them to that will get them towards a diagnosis faster,” he said. “And once they have a diagnosis, get them into a community to find specialists and appropriate care for them given their needs.”

Many of the communities with which the organization works are just emerging from patients, caregivers, or advocates with like interests who are trying to help advance research into a disease and assist patients with a given condition. The Global Genes Foundation Alliance consists of more than 800 different rare disease organizations that span the spectrum, and through those constituencies the organization organizes events and brings people together. The organization not only provides content, but establishes collaborations and partnerships, such as the one with the Rare Disease Diversity Coalition that led to the 2021 RARE Health Equity Summit.

On the issues of equity, diversity, and inclusion, he said, to address those, we must embrace them.

“If we’re going to do this right, we have to live it,” said Martin. That means looking at all the work Global Genes does through the lens of health equity and seeing whether we are being inclusive and diverse.

The Black Women’s Health Imperative launched the Rare Disease Diversity Coalition in 2020. The organization brings together rare disease experts, diversity advocates, and industry leaders to create sustainable solutions to address many of the challenges and barriers faced by rare disease patients of color. Its key goals are reducing disparities in rare disease,
identifying, advocates for evidence-based solutions, and helping achieve greater equality within the rare disease community.

“You’re trying to get a diagnosis. You’re trying to understand what’s going on. You’re trying to get your loved one or yourself the best care possible,” said Linda Blount, president and CEO of the Black Women’s Health Imperative. “In this world, not everybody has the same access. Race and ethnicity matters. Geography matters. Income matters. What you’re exposed to matters.”

She said the Rare Disease Diversity Coalition was created to address these access issues and make sure that things like race, ethnicity, access to information, health insurance, or money don’t stand in the way of someone being able to get a quick diagnosis and quality treatment.

“We’ve got to make our voices known and heard everywhere,” she said.

In discussing action that could be taken to enable solutions that address specific challenges minoritized populations face in the rare disease space, one area that came up was family health history. Martin said in communities of color information often isn’t shared or documented in a way that might be helpful for a caregiver or for a patient to work with their clinician to get to a diagnosis or avoid a misdiagnosis, get access to a trial, or to find a treatment that might be effective for them.

“We want to spend some time digging into this, together, starting by getting out into the communities and that’s where RDDC and the expertise and the relationships that it has are so critical,” said Martin. “And then we need to pull in the rare disease communities, many of whom are struggling with this same issue. How do we be inclusive within our own population, even though there may be only 100 or 1,000 people in our population.”

He said Global Genes will be launching a personal health information management tool, which will have a family health history feature.

To address the wider data gap, Global Genes will be working on the question of how to create the infrastructure to collect information and apply it in an open and collaborative research environment where people can look at that data to better understand rare diseases and how they may affect different communities that are in one way or another minoritized and what the communities may have in common.

Finally, the Rare Disease Diversity Coalition has a clinical trials diversity effort to change the pipeline. Blount noted that National Institutes of Health RO1 funded researchers of color in 30 years hasn’t changed at under 2 percent and the percentage of Black and Latinx physicians hasn’t changed in 40 years.

She said the RDDC’s effort is to change the pipeline, change the way we view evidence, and change clinical trials participation.

“To get to equity, we are going to have to consider a systems change. I’ll quote a very famous philosopher, Moms Mabley, who said, ‘If you do what you’ve always done, you’re going to get what you always got,’” said Blount. “As I said, rare disease patients are fearless. They’re willing to step out. So, let’s do things differently.”

In this world, not everybody has the same access. Race and ethnicity matters. Geography matters. Income matters. What you’re exposed to matters.

— LINDA BLOUNT, BLACK WOMEN’S HEALTH IMPERATIVE
As part of its effort to supporting health equity, diversity, and inclusivity and improve quality care for the rare disease community, Global Genes issued its 2021 Health Equity RARE Patient Impact Grant: Improving Cultural Competency. The grants promote improved outreach to underserved and underrepresented communities supported by RARE Foundation Alliance members.

Global Genes made the awards to groups seeking to expand their research, resource development, outreach, education, and awareness efforts supporting cultural competency. This includes such efforts as establishing a rare disease health equity network of peers, increasing cultural competency across rare disease patient communities through resources and meetings; and developing culturally appropriate communications and materials to help educate minoritized, underserved, and underrepresented patients about rare diseases, diagnostic testing, and the importance of participating in clinical trials. To be eligible for the grant, applicants needed to be members of the Global Genes RARE Foundation Alliance and accredited in the United States.

Grant recipients participated in a series of virtual workshops with the RARE Health Equity Leadership Network and worked with this group to develop ideas to help support equal access to quality care, diagnostics, and research in the rare disease community.


The grants covered a wide range of efforts. In some cases, the grants were used to reach specific underserved communities. The Angioma Alliance used the grant to reach out to better engage Black patients with cerebral cavernous malformation (CCM), while Sick Cells used the grant to conduct research into the unique educational needs for the Hispanic community living with sickle cell disease. In other cases, such as the Foundation for Sarcoidosis Research, programs focused on raising awareness in a more targeted population, such as African American women, who disproportionately suffer from sarcoidosis.
Some organizations, such as the National PKU News, focused on translating resources into different languages. In the case of National PKU News, it combined its three most vital resources and translated it into Spanish to fill a gap that has existed in treatment equity for people with the rare metabolic condition PKU. In the absence of available resources in Spanish, many clinics create ad-hoc translations of their local materials for patients who speak Spanish, but until now none of the primary reference materials have been available for those patients in their first language.

Still others like the CFC International, which is focused on cardio facio cutaneous—a rare condition that affects the heart, facial features, and skin—took a broader approach. It used the grant to establish health equity programming and strategic initiatives and engaged a consultant to work with it on individual and group listening sessions among caregivers in communities of color.

Some of the organizations found that implementation of their programs was harder than they expected. The Angioma Alliance found difficulty identifying and connecting with Black CCM patients. It added 15 Black patients to its patient registry through the program. While it said this was far more than would have been added without the program, it fell short of its expectations.

Its work has led to plans for a public-private hospital partnership because it learned that because of insurance, some of the patients from its community may not have access to the centers of excellence that are hosted in private hospitals. The public-private partnership is intended to offer mentorship to public hospitals by offering programming and consultation focused on the CCM standards of care and the special needs of Black CCM patients.

Sick Cells, in seeking to make available culturally competent materials, found that the level of understanding about sickle cell disease varied within the Hispanic population. It said materials made to educate this community should be made in partnership with Hispanic individuals living with sickle cell disease and go through rounds of focus group reviews to understand what education is needed.

Other groups like CFC International found it had to make choices about priorities. While it initially thought it could do the work in conjunction with other work it was doing, it found half-way through the process that it needed to work with partners in the rare disease space who are focused on health equity.

The Foundation for Sarcoidosis Research, as part of its plan, created a Women of Color Patient Committee to ensure its campaign resonated with patients it sought to target. It said the committee underscored the importance of incorporating targeted communities in the planning and execution of programs. There were several instances where the patient committee identified blind spots that existed in its messaging which helped it avoid pitfalls like using language that could be easily misunderstood or perceived as offensive to the black community of women it is targeting.

Uplifting Athletes, which used the grant to grow the underrepresented researchers in medicine initiative, said it learned it needed to conduct earlier outreach to ensure underrepresented researchers had adequate time to apply for the grant. It is exploring new funding options for a grant for underrepresented researchers in conjunction with a matching grant from a patient advocacy organization.
Promoting Financial Advocacy Among Rare Disease Organizations

Preventing and mitigating the impact of financial hardship can greatly improve care satisfaction, clinical outcomes and quality of life. By helping rare disease patients understand and navigate the healthcare system, we can prevent financial difficulties such as bankruptcy, borrowing, loss of work, as well as financial distress that may come as a result of unexpected barriers to healthcare and support services.

Global Genes’ Financial Advocacy RARE Patient Impact Grant promotes financial health and positive financial decision making along the healthcare journey for RARE Foundation Alliance members.

The one-time grant opportunity was awarded to members of the Global Genes Foundation Alliance based in the United States who were interested in expanding their research, resource development, outreach, education, and awareness efforts supporting financial literacy as well as financial advocacy and planning in healthcare.

Global Genes awarded eight 2021 Financial Advocacy in RARE (F.A.I.R.) RARE Patient Impact Grants ranging from $8,000 to $3,800. The recipients included the E.We Foundation, Akari Foundation, ALD Connect, DDX3X Foundation, RareKC Foundation, United Mitochondrial Disease Foundation, Angelman Syndrome Foundation, and the Dup15q Alliance Foundation.

The grants were used for a wide range of projects. In some cases, these projects focused on narrow needs of specific patient populations. For instance, the Dup15q Alliance provided education around the Special Needs Trust to preserve current and future eligibility for Medicaid and Supplemental Security Income.

Other groups found their members had a broader range of financial issues that needed to be addressed. ALD Connect did broader education to help families navigate state and federal programs and access financial resources.
"We have seen parents and patients in a wide variety of situations, ranging from no insurance to liquidating their retirement to pay for treatment. Some families do not understand the basics of insurance, others try to fundraise to cover expenses, and a few manage to do okay," the organization said. The grant allowed ALD Connect to start financial advocacy projects that it said would not otherwise have happened this year.

Because of the differences in insurance depending on the state someone lives in, the Angelman Syndrome Foundation used its grant to provide state-by-state guides with plans to update them every six months. Similarly, the United Mitochondrial Disease Foundation worked on a series of state specific financial guides with a generic guide for people living in a state that had not yet created a specific guide.

Other groups took a broader approach. RareKC’s Patient Navigator Program, which provides resources and guidance across many aspects of a patient’s life and journey including family financial decisions, medical costs and insurance, educational opportunities, housing, and employment (for both parents and individuals with a rare disease), used the grant to create a first-of-its-kind comprehensive manual addressing the specific needs of patients, families, and caregivers impacted by rare diseases.

The Akari Foundations work was focused on helping low-income Hispanic and immigrant families who are carriers of Duchenne muscular dystrophy or are newly diagnosed through services, information, and educational material in Spanish. Among the work it did was create eight educational webinars in Spanish for Hispanic families.

For some organizations, finding experts who were able to help at times proved challenging. Many experts were concerned about addressing topics that were too expansive and felt unable to address disease-specific concerns.

Groups also found that many people are private about their finances and not comfortable talking about them in public settings.

"We learned that this is a challenging topic to cover because it is so dependent on individual circumstances," ALD Connect said. "We also learned that some families are reluctant to talk about their financial situation, making it more difficult to understand specific challenges. We were not surprised to find that the best source of information is families themselves. Their firsthand knowledge and experience are invaluable."
Connecting Patients to Medical Students to Drive Compassion

As part of Global Genes efforts to support health equity, diversity, and inclusivity and improve the quality care for the rare disease community, the organization established the RARE Compassion Program.

Each year, the program connects U.S.-based medical students with rare disease patients and families to help foster meaningful doctor-patient relationships, inspire careers in rare disease care and research, and ultimately, through patient-centric listening and interactions, build the next generation of future medical professionals as key advocates for their patients.

Medical professionals often don’t receive training to recognize a patient with a rare disease, even though in the United States an estimated 1 in 10 people are living with such a condition. What they know may be limited to what they have read in textbooks, and that may offer a one-dimensional view of what patients with these conditions must go through. The program is intended to foster compassion and awareness for the challenges that patients diagnosed with a rare disease are faced with every day.

For the 2021 RARE Compassion Program, students were paired with two patients over the course of eight months (four months with each patient/patient family). Students and patients were paired based on commonalities related to disease/disease interest. In order to remain active in this program, students and patients were required to meet via phone or video
conferencing once a month for a minimum of one hour for the entire eight-month program.

A total of 294 patients applied to the program, 218 of whom were matched. Of those, 206 patients completed the program. Patients who participated in the program said they enjoyed helping medical students understand the human side of rare disease and appreciated someone wanting to listen to their story and learn from their experiences.

Patients and their caregivers discussed the impact these conditions can have on daily life. One parent of a child who has a rare disorder that is not visible but causes the child to behave differently at times discussed the way people’s lack of understanding impacts their lives.

“We discussed how hard it is to do simple daily things like going to the store without a potential outburst or her running away leading to a scene and stares of disdain from other people. We talked about having a support group and that having a close group of accepting friends has really improved our emotional health and allows all of our children to play without having that feeling like people are staring at us,” the parent said. “Our discussion also included how our entire family is impacted and the relationships that can be strained from being burnt out and stressed. The toll on our own mental and physical health to care for a child with a rare disorder was also discussed.”

A total of 120 students applied to the program, 108 of whom completed it. These students collectively spent 463 hours meeting with patients through the program. These students said the experience gave them a better understanding of the patient-child relationship in navigating rare diseases, the importance of language in coloring patient interactions, and an understanding of the challenges patients face from navigating insurance to finding the right doctor.

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“Up until this point in my academic and clinical career, most of my exposure and understanding of rare disease has come from textbooks and lecture videos. To be able to actually discuss with a patient and his family the implication and impact of a rare disease diagnosis was one of the most eye-opening and substantial experiences I have had in terms of my medical education. I got an introduction to the many aspects of rare disease and the many ways it affects patients and their families.”

Students who participated in the program are eligible to apply for Global Genes’ David R. Cox Scholarship for RARE Compassion grants.
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