mediaplanet

Rare Diseases

February 2025 | www.futureofpersonalhealth.com

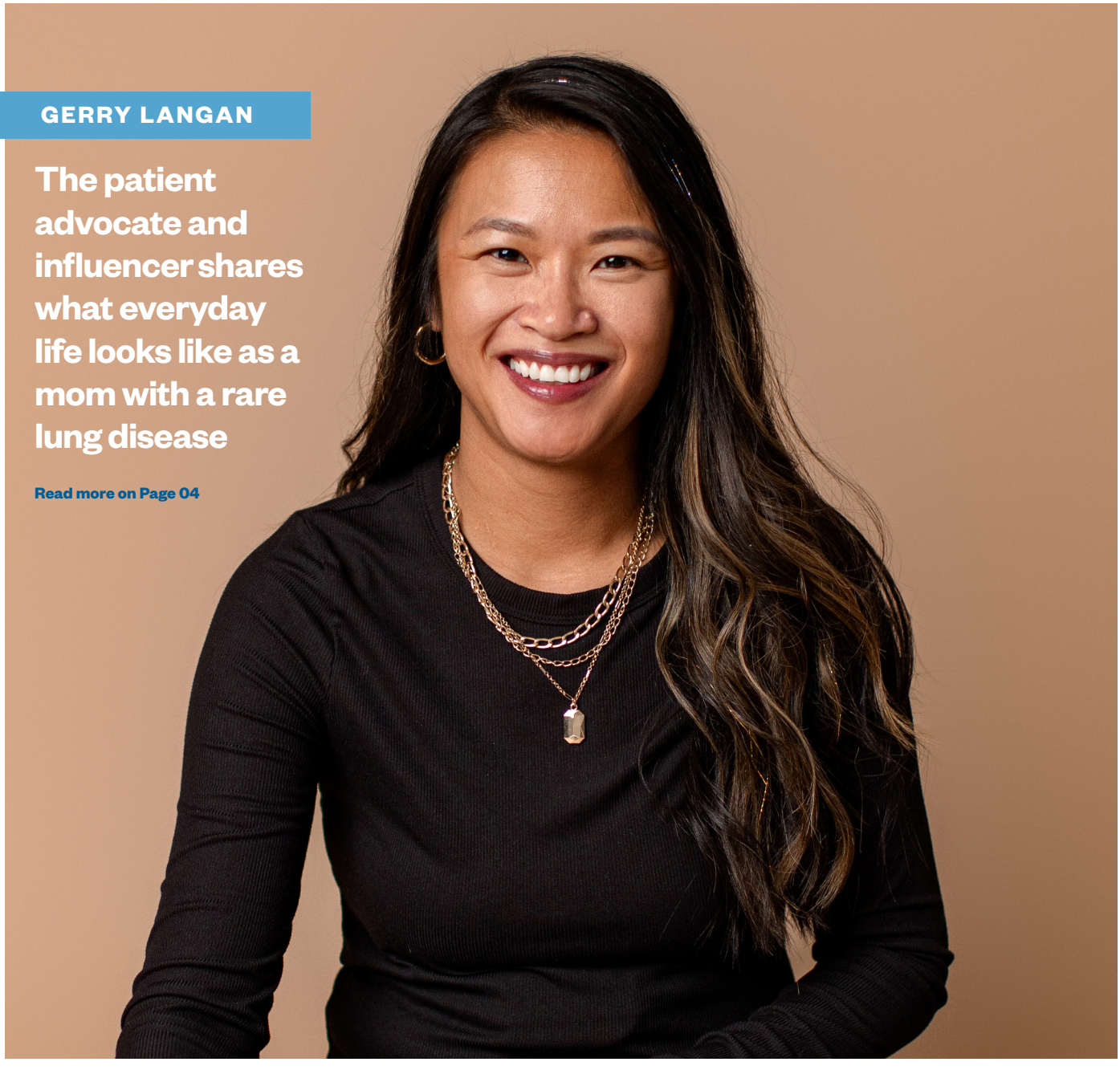
An independent supplement by Mediaplanet to USA Today

"Understanding a rare disease, even without a cure or treatment, is incredibly important."
Dr. Tomi Pastinen, Director, Genomic Medicine Center, Children's Mercy Kansas City

Page 03

The introduction of new therapies gives hope to the millions of Americans living with rare disorders.
Drs. Lindsay Burrage and Jerry Vockley, American College of Medical Genetics and Genomics

Page 06



GERRY LANGAN

The patient advocate and influencer shares what everyday life looks like as a mom with a rare lung disease

Read more on Page 04

Shortening the Diagnostic Odyssey **Through Newborn Screening**

For over 50 years, a simple heel prick has detected disorders in newborns that, if left untreated, cause debilitating illness or death, making early diagnosis critical.

Newborn screening programs test every newborn for genetic, metabolic, hormonal, and functional conditions that are not otherwise apparent at birth. Each year, approximately 4 million U.S. newborns are screened, and about 1 in 300 are identified to have a potentially devastating condition, benefiting from early detection and the delivery of life-saving treatments.

Tragically, however, our nation's current newborn screening system is unsustainable, and our babies are at risk. The federal law that supports the national newborn screening program expired two years ago. Additionally, there are life-altering discrepancies between which conditions are screened for in each state.

The EveryLife Foundation for Rare Diseases is leading the fight to ensure that the newborn screening program is prioritized so that families receive timely diagnoses and early interventions. These laws require all states to screen newborn babies for any disorder on the federal Recommended Uniform Screening Panel (RUSP), implement a timeline in which the screening must begin, and ensure that resources will be available to fund all conditions added to the RUSP in the future.

Written by **EveryLife Foundation for Rare Diseases**

Leveraging the Power of Model Organisms to Diagnose and **Develop New Therapies for Rare Diseases**

Studies of human disease-causing genes can often be performed in model organisms like yeast, fruit flies, and mice, leading to breakthroughs in diagnoses and treatment.

It is estimated that there are more than 10,000 rare diseases affecting more than 20 million people in the United States and 400 million worldwide. An estimated 80% of these rare diseases are caused by mutations in our genes.

Prior to 2010, it was very difficult to pinpoint the specific genes and genetic mutations that cause rare human diseases. However, extraordinary advancements in technology have completely changed the landscape of human genetics, and today, scientists can sequence the entire human genome for less than $1,000. By sequencing the genomes of individuals who have rare diseases, geneticists are able to identify genetic variants that could be causing disease, which are then studied in follow-up experiments.

The experimental power of model organisms

All over the world, scientists are capitalizing on new technologies to advance discovery for rare genetic diseases. By using the experimental power of model organisms, scientists can conduct studies to determine the genetic basis of disease and help provide individuals with a diagnosis for the first time.

Functional studies of human disease-causing genes can often be performed in genetic model organisms, such as yeast, worms, fruit flies, zebrafish, and mice/rats. Indeed, a comparison of the genomic structures between these organisms and humans reveals that all organisms are built from a similar set of genes. For example, fruit flies share 75% of human disease-causing genes. Thus, experimental manipulations of model organisms can be used to understand the biological effects of a disease-causing mutation. This approach allows scientists to determine whether, and how, the human genetic variants affect the normal function of the gene.

Unlocking new therapeutic approaches

While a precise DNA molecular diagnosis provides some relief to patients who have undergone a diagnostic odyssey, it is just the beginning. Identifying the cellular and biochemical pathways that are at the root of a patient's symptoms is the most straightforward way to develop potential new therapies. In these endeavors, the model organisms provide unmatched tools to dissect the disease mechanisms.

By rapidly assessing the precise expression pattern of the genes, the dynamics of the proteins they encode, and the affected biological pathway, researchers can identify potential therapeutic targets. Furthermore, by unraveling the molecular players in the disease, scientists can, in some cases, identify FDA-approved drugs that can be repurposed to target the molecular pathway. These discoveries have already led to new therapeutic approaches, and physicians have applied for compassionate use of drugs when appropriate for their patients.

Going forward, it is essential to maintain robust levels of research funding to support scientists' functional studies on the genetic causes of disease using model organisms. These critical discoveries can then be translated into new therapeutic strategies and clinical studies to help as many patients as possible. Continuing to expand connections, collaborations, and crosstalk between basic scientists, clinicians, and patients will also be key in advancing discovery for rare genetic diseases.

Written by **Hugo J. Bellen and Philip Hieter, Genetics Society of America**

@futureofpersonalhealth @MediaplanetUSA Contact information: **US.editorial@mediaplanet.com** Please recycle

Publisher **Shaina Heitzner** Managing Director **Gretchen Pancak** Production Manager **Dustin Brennan** Creative Director **Kylie Armishaw** Director of Client Success **Taylor Daniels** Designer **Celia Hazard** Copy Editor **Taylor Rice** Cover Photo by **Grace Oliver Photography and Victoria Chandler Photography** All photos are credited to Getty Images unless otherwise specified. This section was created by Mediaplanet and did not involve USA Today.

Unlocking Answers: Transforming Rare Disease Diagnosis

Imagine waiting years to find out what is wrong with your child. For families dealing with rare disease, this is a harsh reality, and the journey is often long and filled with uncertainty. On average, it takes about 4-5 years for a child to be diagnosed, but for some, the wait is even longer. Even more startling, two-thirds of children with rare diseases remain undiagnosed.

That is why Children's Mercy Kansas City launched Genomic Answers for Kids (GA4K) in 2019. This first-of-its-kind pediatric data repository empowers the search for insights and novel treatments for genetic conditions in children. Our goals are to expand access to genomic testing and develop cutting-edge genomic testing to help more families get answers faster and disseminate the learnings from the new genomic sequencing in rare disease clinical research community to impact patients worldwide.

The heart of GA4K is identifying genetic causes for childhood illness, which are individually rare, but collectively impact 1 in 20 Americans. One of the key tools to finding genetic causes of illness is genome sequencing. Genome sequencing reads the book of a person's genetic information, or DNA, letter by letter. The DNA between any two humans is 99.9% the same. The 0.01% difference between humans is what gives us different characteristics and appearances. Some of these genetic differences can also lead to disease, identification of which allows us to determine cause of childhood disease.

5-base HiFi sequencing

Through the GA4K program, we have pioneered the world's most-advanced genomic sequencing technology, 5-base HiFi sequencing, and we've made incredible progress in understanding the human genome. For the first time, our researchers can now sequence the full genome and methylome in one test.

This means we can access patient DNA all at once, rather than needing multiple tests from different labs, and detect all types of variation in the genome. Thanks to this breakthrough, we can now identify previously undiagnosed mutations that cause rare disease.

GA4K has also paved the way for integrating 5-base HiFi sequencing into clinical care, benefiting more patients beyond research. We were the first in the country to use this technology in a clinical setting, speeding up diagnoses and providing more answers than other tests for patients.

This consolidated test provides efficient diagnosis for kids with rare, complex genetic diseases, and increases opportunities to have all treatment and management options available as early as possible for ill patients in the hospital. Results that used to take months can now be achieved in two weeks with HiFi sequencing.

Better diagnosis, better care

Understanding a rare disease, even without a cure or treatment, is incredibly important. Identifying the cause of a child's symptoms can bring immense relief to families, and it enables care teams to manage symptoms more effectively. A diagnosis can also connect families with support groups, educational resources, and specialized care that might otherwise be inaccessible. Turning uncertainty into knowledge empowers families to navigate the challenges of rare disease with greater confidence and support.

Improving rare disease diagnosis and treatment requires collaboration across children's hospitals, and healthcare institutions and disciplines. Many of the patients enrolled in GA4K are being treated at Children's Mercy, but the program is inclusive and open to children who may have a genetic disease not resolved by standard testing. We have eight partner institutions from around the world currently nominating and enrolling patients, and providers from other institutions can also contact us directly.

Through the groundbreaking and ambitious effort of GA4K, we have made significant strides in the field of pediatric genomics. Children's Mercy has involved over 15,000 patients and family members in GA4K and provided rare disease diagnoses for more than 2,000 families to-date. And we are committed to helping even more children and families find answers.

WRITTEN BY
Tomi Pastinen, M.D., Ph.D.
Director, Genomic Medicine Center, Children's Mercy Kansas City

To learn more, visit
cmkc.link/GenomicA4K

Children's Mercy KANSAS CITY

Health Advocate Gerry Langan on Living a **Normal Life With a Rare Lung Disease**

After being diagnosed with pulmonary arterial hypertension (PAH) and heart failure at 28 years old, Gerry Langan dedicated her life to becoming a patient advocate.

Can you share your journey leading up to your PAH diagnosis and how it impacted your life?

I was diagnosed about a year after I had our twin boys. We had moved from North Carolina to Colorado, which is at a high altitude. Initially, my symptoms looked like altitude sickness or postpartum issues. Isolated, each symptom seemed explainable. However, as they started to compound, it became clear something was really wrong. I was extremely tired, could barely walk, was retaining a lot of fluid, and had trouble breathing. Separately, these things seemed manageable, but together they painted a much bigger picture.

I went to the doctor, thinking I might be pregnant, but I wasn't. Less than a week later, I ended up in the emergency room, where they diagnosed me with heart failure. The heart failure was secondary to PAH. By the time I was diagnosed, my PAH was so advanced that it had caused severe heart failure.

The diagnosis was life-changing. After seeing a specialist, I was rediagnosed and had to start treatment all over again when we moved to a different state three weeks later. Adjusting to a new medical team and treatment plan was challenging. At the time, everything felt so reactive. We didn't have much time to process how extreme the situation was — we just had to keep going.

Gerry Langan

Are there any misconceptions about PAH that you aim to dispel through your content?

Dealing with a rare disease like mine comes with a lot of challenges, especially when it comes to educating people about it. For one thing, my disease is extremely rare. People often come to my page thinking they might have it, and while I try to be sensitive to that possibility, it's important to emphasize how rare it actually is. It's not commonly diagnosed, so in many cases, it might turn out to be something else entirely.

Another source of confusion is pulmonary hypertension (PH). People sometimes confuse it with regular hypertension, which is much more common. I'll get comments like, "Oh, you can just take medication and live a long life," and while that's true for regular hypertension, it's not the case for what I have. My condition, PAH, is a type of PH that's even rarer.

It doesn't help that PH is often used as a catch-all term. Technically, PH is the broader category, with subtypes like PAH. PAH is a pulmonary (lung) disease, not a heart disease, although it can cause heart issues. This leads to confusion. People ask why I'm not seeing a cardiologist when my primary doctor is a pulmonologist, as the root issue lies in my lungs.

Explaining all this can be exhausting: the distinction between heart and lung diseases, the subcategories of PH, and why I use the term PH instead of PAH for simplicity. It's a never-ending cycle of explaining, educating, and trying to live with the daily realities of a rare and misunderstood disease.

What are your hopes for the future?

Right now, my main focus is being present for my kids. They're growing up so fast, and I want to make sure I'm there for them. This disease is progressive, and while I'm doing better thanks to a clinical trial and advancements in medical care, there are no guarantees. I want to cherish every moment with my family.

As for my health, my goal is to be cured. I want to get off the pump, and I know my doctor will probably roll his eyes if he hears this, but we've been working toward that goal. Unfortunately, it's not a reality right now, and it was a bit disappointing when I got that news a few weeks ago. But I have to remind myself that I feel really good overall and am living a nearly normal life — minus the doctor's appointments and treatments, of course. I'm thankful for that.

Why We Advocate: The Importance of Access to Efficient, High-Quality Healthcare

Anyone living with a rare disease wants access to care — not just substandard healthcare, but healthcare that will keep them alive and healthy. Sadly, this is often not the case for so many patients.

I am Laura Bonnell, the mother of two daughters with cystic fibrosis. For 30 years, I have advocated for my daughters to ensure they get the care they deserve, and to promote legislation that will assure their basic rights to care.

A couple of years ago, when my daughter Emily was 25 years old, she had a horrible lung infection. I drove her to the emergency room, where she was admitted right away. She went into surgery, and the PICC line was placed in her arm so she could do intravenous (IV) medication for about 6 weeks at home. On our way home, we had to stop at the pharmacy to pick up her oral medication, linezolid.

We called ahead to our local pharmacy, and the pharmacist said they were out of the medication. We asked if they could find it for us at another pharmacy because we only had a one-hour window before we had to be home to do her IV dose of medications. The pharmacist changed her story and said they actually did have the pill, but they couldn't give it to us.

We were confused and out of time, so we drove to the pharmacy anyway. Emily was pale and exhausted. When we were standing before the pharmacist, she said the real reason she couldn't give it to us was because we didn't have prior authorization. Prior authorization is needed before your insurance will pay for certain medications. The hospital discharge nurse assured us before we left that they had taken care of this, but somewhere in the process, things got messed up. The hospital pharmacy was closed, and this meant we couldn't fix the problem until the next day. Emily couldn't wait that long.

The role of patient advocacy

At this point, Emily was not even close to turning a corner. I said to the pharmacist, "If you're not going to give us the medications, I am going to need you to take all the signs down that say things like, 'We're here to help.' We are caught in the middle by an insurance situation that is not our fault. We are covered by not one, but two health insurance companies, and we're not leaving until we get her medications."

At this point, the pharmacist was frazzled, Emily was crying, and I was desperate. "I am not leaving without her medication," I said. "Please give us three pills so we can make it until tomorrow. Look at my daughter — please do not compromise her health."

After a minute of thinking, the pharmacist quickly threw three pills in a prescription bottle and handed it to me. She didn't say a word.

When we got home, I realized she put the pills in an unmarked prescription bottle — no linezolid label. I told Emily that if these pills didn't have the name written on them, she couldn't take them. Fortunately, when I opened the prescription bottle, I saw the name of the medication written on the pill. The entire situation was scary and unnecessary. This is why we advocate.

WRITTEN BY
Laura Bonnell
Founder, The Bonnell Foundation

Don't Ignore the Signs: Rare Diseases Are More Common Than You Think

Rare diseases aren't as rare as you might think. An estimated 1 in 10 Americans has one, and more than 50% are children, many still undiagnosed.

On average, it takes five years to get an accurate rare disease diagnosis. That's five years of unanswered questions, misdiagnoses, and missed opportunities for early interventions.

The National Organization for Rare Disorders (NORD®) is hoping to reduce the diagnostic journey by sharing steps to take when your child or a child you care for has unresolved health issues:

1. Trust your gut

No one knows your child better than you do. If something feels off, listen to that inner voice. Parents and caregivers often recognize a problem long before it's identified medically.

2. Partner with your physician

Your child's pediatrician should be your first stop. Bring them a detailed symptom log, and ask questions about potential underlying conditions. If your concerns aren't being addressed or you don't feel heard, seek a second opinion.

3. Educate yourself

If you or your child receives a rare disease diagnosis, the NORD® Rare Disease Database offers information on over 1,400 rare conditions, treatment options, and clinical trials. Understanding the disease will help you make informed decisions about your child's care.

4. Participate in research

Fewer than 5% of the 10,000+ rare diseases have Food and Drug Administration-approved treatments. New treatments rely on families participating in clinical trials, registries, and genetic studies.

5. Don't give up

The rare disease journey can be long and uncertain. Yet, progress is happening every day. With persistence and support from organizations like NORD®, answers and help are possible.

Written by **National Organization for Rare Disorders Disorders (NORD®)**

Paving the Way to Better Health: **A Guide to Clinical Trials for Rare Diseases**

By working together with patients, doctors and researchers can learn more about rare diseases and improve outcomes through clinical trials.

Why is it important for patients with rare diseases to join clinical trials?

Without clinical trials, treatments would be limited or non-existent for rare diseases. Participating in trials helps researchers gather the data needed to better understand the disease and develop effective treatments. By joining clinical trials, it will also ensure your experiences and needs are considered. Patients in clinical trials often receive specialized attention from medical experts and access to advanced diagnostics, which can improve their overall care.

How can clinical trials impact patients' lives?

Patients gain access to experimental treatments that might not be available elsewhere, most likely improving their condition or quality of life. Clinical trials usually include advanced monitoring and testing, which can help patients and their doctors better understand the progression and management of their specific condition. There may also be an opportunity to meet with other patients to feel less alone.

How can patients find and join clinical trials?

ClinicalTrials.gov is a global database listing thousands of clinical trials across diseases, including rare conditions. Patients can search by disease, location, or treatment type.

Written by **Ellyn Kodroff, President, Campaign Urging Research for Eosinophilic Disease (CURED)**

New Therapies Offer Hope for **People with Rare Diseases**

New advances in medical technology have improved how we identify rare genetic disorders and how we create treatments for them.

Rare genetic disorders are caused by changes, or variants, in DNA. Variants in DNA can lead to problems with making RNA or proteins, which then can cause genetic disorders. New tools, like DNA sequencing, have made it easier to diagnose genetic disorders quickly. Because of these tools, researchers and doctors can now focus more on developing new treatments.

Phenylketonuria (PKU) is a good example of how genetic disorders can be treated. PKU happens when there are variants in the gene that make a protein called phenylalanine hydroxylase (PAH). In people with PKU, the PAH protein does not work correctly, so it cannot turn the amino acid phenylalanine into another amino acid called tyrosine. This causes phenylalanine to build up in the blood, leading to brain damage and severe intellectual disabilities if not treated.

Luckily, if PKU is found early, it can be treated with a special diet that limits phenylalanine. This helps prevent brain damage and allows for normal development. PKU is one of many disorders that can be detected in newborns before symptoms start. Early diagnosis and treatment have greatly improved outcomes for people with PKU. However, the special diet can be expensive and hard to follow.

Making treatment options accessible

New treatments are giving hope to people with PKU. One treatment helps the PAH protein work better. Another drug uses a different protein to lower phenylalanine levels in the blood. Scientists are also working on treatments to help the body produce normal PAH protein. These new treatments include RNA therapy and gene therapy. RNA therapy is a temporary fix that needs to be repeated over time because RNA breaks down in the cells. On the other hand, gene therapy can offer a long-term cure. Gene therapy is now becoming a reality, with new gene therapies approved for almost 2 dozen disorders.

Unfortunately, these new treatments are very expensive. Developing drugs for rare disorders costs a lot of money because only a small number of people need them. As a result, the prices for these treatments can be hundreds of thousands or even millions of dollars. Rapid advances in diagnosis and the introduction of new therapies give hope to the millions of Americans living with rare disorders and to the physicians caring for them. Moving forward, doctors, patients, payers, and researchers will need to work together to make sure that available treatments are accessible to all who need them.

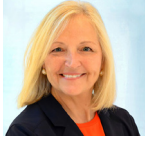
Written by **Lindsay C. Burrage, M.D., Ph.D., FACMG, Chair, Therapeutics Committee, and Jerry Vockley, M.D., Ph.D., FACMG, Board of Directors, American College of Medical Genetics and Genomics**

Bridging The Gaps in Rare Disease

Advocacy and Support

Our panel of experts discusses the multifaceted obstacles in rare disease advocacy and how collective efforts can drive rare disease awareness, support, and research.

INTERVIEW WITH
Christina Suh, M.D., M.P.H
Director of Clinical Affairs, Phreesia



INTERVIEW WITH
Pamela K. Gavin
CEO, National Organization for Rare Disorders (NORD ®)

INTERVIEW WITH
Michael Eging
Executive Director, Rare Access Action Project (RAAP)

What are the biggest challenges rare disease advocates face in raising awareness among policymakers, healthcare providers, and the general public?

Christina Suh: One of the biggest challenges rare disease advocates face is the timing pressure that today's clinicians are up against. Healthcare providers are repeatedly told "When you hear hoofbeats, think horses, not zebras," in part because, in many situations, it helps us reach the right diagnosis faster. If a patient needs a specialty follow-up, the provider must find time for a referral, a process that still often relies on fax machines. If genetic testing is recommended, that takes time to coordinate.

Technology can save time at these critical steps. Rare disease advocates can better support families by helping providers co-create new technologies that address the realities that providers face.

Pamela Gavin: Additionally, few rare diseases are understood and studied by the medical community, and most lack sufficient research funding. This lack of knowledge among healthcare and insurance providers leads to delays in diagnosis and treatment, which the right policies can help address. Despite progress, fewer than 5% of rare diseases have a Food and Drug Administration-approved treatment, and over 200 new rare diseases are discovered annually, outpacing drug development. This is why incentives for drug development are so important.

What are the most pressing gaps in support services, and how can stakeholders work together to address them?

PG: A major gap in rare disease support is the financial burden on families. The excessive cost of treatments, prescriptions, and routine care forces families to make impossible choices, like rationing medications, quitting jobs, or forgoing care. This strain harms both the patient and their loved ones, impacting health and quality of life.

Michael Eging: The healthcare system throws up continued barriers to access to both care and therapies, leading patients to enormous frustration. Exploring issues that will reduce those barriers as well as lower out-of-pocket costs would significantly improve the well-being of rare disease patients.

What gives you hope for the future of rare disease advocacy, support, and research?

ME: Rare disease patients are persistent, and increasingly they are becoming very savvy with various methods of communication needed to educate and advocate stakeholders and policymakers. The rising generation of rare disease advocates has been effective in opening doors to generate policy solutions and educate policy stakeholders on the negative impacts of policies such as copay accumulators, PDABs, and Medigap issuance for patients under the age of 65. The policy advancements rare disease advocates have provided have shined a light on bad policy, promoted workable solutions, and provided hope for rare disease patients that the therapies and coverage they need will be provided.

CS: We're seeing a real shift toward person-centered care, and I'm encouraged by the ways in which patients can and do advocate for themselves. Patient activation for the general population is more important than ever when it comes to rare diseases because more activated patients can get a diagnosis faster. This, in turn, makes it possible to treat the condition when it's still possible to see good outcomes. This self-advocacy is not only raising awareness of rare diseases but also driving change in research and policy.

To learn more about Rare Diseases,
visit **futureofpersonalhealth.com**

mediaplanet