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Priya Kishnani, Director of the Division of Medical Genetics at Duke University School of Medicine, discusses how multidisciplinary care, global collaboration, and rapidly advancing therapeutic technologies are reshaping the rare disease landscape. Drawing on decades of experience across diagnosis, natural history research, and the development of transformative treatments, she outlines the progress made in the field while addressing the remaining challenges around access, clinical trial design, and equitable delivery of innovation to patients worldwide.

Could you briefly introduce the overarching responsibility of the division of medical genetics and the extend of which rare diseases fit into that scope?

The division of medical genetics sits within the department of pediatrics, and we are a highly multidisciplinary group that brings together clinicians, laboratory experts, and researchers. Many of our faculty are certified in both clinical and biochemical genetics, which allows us to care for individuals with genetic and metabolic conditions across the entire lifespan from newborns through adulthood and into older age. Although we are housed in pediatrics, our clinical reach extends well beyond it.

A key strength of the division is our integrated diagnostic laboratory. While the lab operates within the hospital, the PhD faculty who direct it are members of our division, which allows for very close collaboration and rapid turnaround of results. We house advanced diagnostic capabilities, including

mass spectrometry and enzymology. We also run the Glycogen Storage Disease (GSD) Lab, which is the only enzymology lab of its kind in the US and one of very few worldwide.

In parallel, the division has strong clinical, translational, and bench research programs. Several faculty are members of the Undiagnosed Diseases Network, and others have played central roles in the development of therapies for rare diseases. This includes work on enzyme replacement therapy for Pompe disease, gene therapies for Pompe and other glycogen storage diseases, and siRNA-based approaches for metabolic disorders such as glutaric acidemia type I. Our research spans enzyme therapies, AAV-based gene therapies, gene editing technologies including prime editing, and detailed natural history studies supported by both clinical programs and animal models.

Collaboration is central to how we operate. We work closely not only across pediatrics but across the broader campus, including engineering and other scientific disciplines, to fully leverage institutional expertise for patients with rare diseases. The division also houses the Chen Center, which supports bench-to-bedside discovery for single-gene disorders, and we are recognized as a NORD Center of Excellence. Together, these elements allow us to deliver comprehensive care and cutting-edge innovation for patients living with rare genetic conditions.

How important is multidisciplinary care across specialties and externally with other hospitals or academic centers in ensuring comprehensive care for rare disease patients?

Multidisciplinary care is absolutely essential, and it operates at multiple levels. There is multidisciplinary care at the point of direct patient care and also in research. Both are critical for rare disease patients.

From a clinical perspective, when a patient with a rare disease comes to us, we try to make that visit as comprehensive as possible. It is not just about convenience, although that matters, but about maximizing expertise and making sure we are addressing all aspects of the disease. For example, in muscle disorders, you are not just dealing with skeletal muscle. There are cardiac implications, smooth muscle involvement, pulmonary considerations, neurological aspects, and biochemical markers that all need to be evaluated. Each rare disease has its own set of needs, and without input from multiple specialties, you risk missing important pieces of the clinical picture.

This multidisciplinary approach is also essential for advancing our understanding of disease. Without these different perspectives, we not only compromise patient care, but we also limit our

ability to fully understand disease progression and natural history.

Beyond internal collaboration, external partnerships with other hospitals and academic centers are equally important. Many patients are referred to us from elsewhere, and maintaining strong communication with their local care teams allows for continuity and shared learning. These collaborations often extend beyond the individual patient and lead to broader data sharing, including natural history studies, registries, and biobanks. With appropriate consent, samples collected locally can contribute to centralized research efforts, and vice versa.

The benefit of this kind of collaboration is enormous. It improves patient care on an individual level, but it also accelerates scientific understanding and therapeutic development.

To what extent do you consider collaboration between clinicians, researchers, and industry to be an important aspect for advancing treatment innovation and outcomes for rare diseases?

None of us can work in silos. Whether you call it a trifecta or a multi-stakeholder model, the reality is that the more perspectives and expertise you bring together, the more progress you can make. Clinicians, policymakers, regulators, industry, and researchers all play distinct but complementary roles, and at the center of it all is the patient. Without strong collaboration and open crosstalk, it is very difficult to move the field forward.

I am a strong believer in this approach, and it is reflected in the way we work. Most of our publications and research efforts involve collaboration not only across the US, but internationally as well. One example is our work on understanding immune responses to therapeutic proteins. In the context of Pompe disease, we partnered with physicians around the world and provided a standardized protocol for initiating enzyme replacement therapy. We then collected and analyzed data across sites and, in collaboration with industry, took those findings to the FDA as part of a post-marketing commitment. That work ultimately contributed to a change in the drug label for alglucosidase alfa and helped establish a new standard of care.

We have seen similar benefits from collaboration in natural history studies. For example, in a rare muscle disease caused by GSD9A1, we worked with clinicians globally to pool data and better understand disease progression. That shared knowledge allowed us to define clinical patterns more clearly and move closer to improved management and treatment strategies.

Collaboration has also been critical in advancing in utero therapies. Through a partnership with UCSF, led by Tippi MacKenzie, and with Duke serving as a site, we explored prenatal enzyme therapy for lysosomal storage disorders. The first patient treated during the COVID-19 pandemic, outside the original trial, was reported in *The New England Journal of Medicine*. Since then, additional patients with disorders such as MPS I, MPS II, and neuronopathic Gaucher disease have been treated, demonstrating proof of concept and opening the door to broader applications, including gene and small-molecule therapies.

We have applied the same collaborative model to hepatic glycogen storage diseases. In conditions such as GSD IX gamma 2, we developed animal models locally but relied on global clinical data to confirm disease progression in patients, including the risk of end-stage liver disease. That collective effort allowed us to draw conclusions that would not have been possible at a single center.

In rare diseases, no one institution or stakeholder can do this alone. Progress depends on shared data, shared expertise, and shared commitment. When we work together across disciplines and borders, we can truly change outcomes for patients.

How do you see new therapeutic technologies reshaping clinical practice and patient care in rare diseases?

There are many new technologies and modalities that are truly lifesaving. While some are more advanced than others, the impact across the field has been remarkable. For example, we are already seeing approved antisense oligonucleotide (ASO) therapies and siRNA-based treatments changing outcomes for patients. The work coming out of players like Alnylam Pharmaceuticals has transformed care for patients with conditions such as porphyria and amyloidosis.

Enzyme replacement therapies have also evolved significantly, extending well beyond lysosomal storage disorders into areas like rare bone diseases, with hypophosphatasia being a clear example. At the same time, small-molecule therapies are now available for conditions such as achondroplasia, which would have been unimaginable not long ago.

Gene therapy has also been particularly transformative. AAV-based approaches have already proven lifesaving in spinal muscular atrophy (SMA) and are now being applied in diseases such as Duchenne muscular dystrophy, with many additional programs advancing through development.

Together these innovations signal just how much the field has advanced.

What are the key challenges that still need to be overcome from diagnosis, access, clinical training for rare disease innovations to reach all patients in need?

Several challenges still need to be addressed to ensure these innovations can truly reach their full impact potential. The first is diagnostic delay and the long diagnostic odyssey that many patients still face. We have made meaningful progress through newborn screening for certain rare diseases which has been completely transformative. We now recognize that days matter. Early diagnosis is critical for lethal conditions and, for others, it prevents years of uncertainty and enables earlier management and timely intervention. Expanding and optimizing these screening approaches remains essential.

The second challenge is our limited understanding of disease natural history. Robust natural history data are foundational for drug development, yet they are particularly difficult to generate in ultra-rare diseases or in conditions with highly variable and complex presentations. Without this knowledge, it becomes much harder to design effective trials or measure meaningful outcomes.

A third challenge is the identification of appropriate clinical trial endpoints and biomarkers. These are essential not only for regulatory approval, but also for guiding real-world clinical care. We still lack reliable markers for many rare diseases that accurately reflect disease progression or treatment response.

Finally, clinical trials themselves capture only a very narrow slice of the patient population. Strict inclusion and exclusion criteria, limited geographic reach, and small sample sizes mean that many patients are excluded. This is why global collaboration is so important. We need to work across institutions, countries, and healthcare systems to pool data, share expertise, and ensure that trials and therapies reflect the full diversity of patients affected by rare diseases.

Addressing these challenges will require continued collaboration between clinicians, researchers, policymakers, and industry, all working together with patients at the center. Together we can make sure that advances in rare disease science translate into equitable access and meaningful outcomes for all patients.

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