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> We are probably living through the most exciting moment in the history of genetic medicine

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Dr Carlos Prada, Director of the Edwards Family Division of Genetics and Rare Diseases at Ann & Robert H. Lurie Childrenâ??s Hospital of Chicago, discusses how earlier diagnosis, coordinated care pathways, and emerging therapeutic modalities are reshaping outcomes for children and families. From newborn screening and multidisciplinary follow-up to the realities of clinical trials and next-generation genetic therapies, Prada highlights both the progress being made and the system-wide alignment still needed to translate innovation into everyday care.

To begin, could you briefly introduce your role at Lurie Childrenâ??s Hospital and outline the scope of responsibility of the Edwards Family Division of Genetics and Rare Diseases?

One of the unique things about genetics is that we really touch almost every specialty across the hospital. Therefore, the Division of Genetics and Rare Diseases is a very multidisciplinary group. We work closely with teams in the NICU, cardiology, nephrology, hepatology, and many others, because rare diseases can present in so many different ways and affect nearly any body system.

At the heart of what we do is helping families get answers, ideally as early as possible. Newborn screening is a great example of how early diagnosis can make a real difference by identifying conditions soon after birth. We try to apply that same approach when children come to the hospital with symptoms that are unexplained or difficult to diagnose. While any one rare disease may be uncommon, when you look at them together, rare diseases are actually quite common. That is why using genetic testing early in the diagnostic process is becoming increasingly important.

Many hospitals are moving in this direction, but it takes more than just ordering tests. You need the right infrastructure in place. That includes access to genetic testing, as well as genetic counselors and care teams who can sit down with families and walk them through the results. Families are often already under a lot of stress, and part of our role is helping them understand how genetic testing can bring clarity and guide next steps.

Sometimes a diagnosis leads directly to a treatment. Other times, even if there is not a specific medication available yet, having an answer still matters. It allows us to put the right supports in place, plan care more effectively, and, in many cases, improve outcomes and quality of life for both the child and the family.

Once a newborn is diagnosed with a rare or genetic condition, what does the clinical pathway typically look like, and how is care coordinated to ensure timely follow-up and long-term management?

Newborn screening is coordinated at the state level, and every baby is screened about 24 hours after birth. Those results are typically sent back to the childâ??s pediatrician who will reach out should a screening result comes back abnormal, that is usually how we first get involved.

Because these are screening tests and not diagnostic tests, the first step is making sure families get information quickly and clearly. We often use telemedicine at that stage, especially since families are at home with a newborn. It allows us to explain what the result means and what the next steps are without delay. We then move quickly to confirmatory testing. In many cases, the screening result turns out to be a false positive, so confirmatory testing helps us either rule a condition out or confirm a diagnosis.

If a diagnosis is confirmed, we bring the family into clinic as soon as possible to talk through treatment and long-term care. What that looks like varies widely. Sometimes requires lifelong dietary changes that need the guidance of a specialized team. In other cases, it may involve specialty medications or infusion-based therapies. As more conditions are added to newborn screening and more treatments become available, the complexity and volume of care continue to grow.

Newborn screening itself is also evolving. Across the country, there are pilots underway to expand screening beyond traditional methods like mass spectrometry and move toward next-generation sequencing. That shift could expand screening from roughly 60 or 70 conditions to potentially hundreds. But identifying more patients also means hospitals need to build the right infrastructure to support them.

At Lurie, we have spent the past few years building that capacity. We now have a dedicated inpatient team focused on newborn screening and metabolic disorders, because as more patients are identified, more children also require hospital admission and specialized care early on. That includes aligning across departments and making sure we have the right expertise in place. For example, some treatments rely heavily on specialized diets. That means working closely with registered dietitians who understand genetic and metabolic conditions and who can help families access

specialized formulas that are prescribed and not available in regular stores. There are many moving pieces, and coordinating all of them is essential to delivering effective care.

What makes this work especially meaningful is that early diagnosis can completely change a childâ??s trajectory. These are children who might otherwise develop serious complications, and with timely intervention, we can often prevent that. As genetic medicine continues to advance and new therapies become possible, we are able to help more patients than ever before.

How is Lurie Childrenâ??s engaging with emerging modalities such as gene and RNA-based therapies, and how prepared is the current clinical ecosystem to adopt and deliver these innovations?

As an academic institution, one of our core missions is to help bring more treatments into the clinic. That really starts with two priorities: making diagnoses earlier and helping move new therapies from development into real-world care. For emerging approaches like gene and RNA-based therapies, clinical trials are the key bridge between those two goals.

To do that well, we need to be very efficient as a clinical trial site. As soon as we know a trial is coming, we start thinking about how to identify eligible patients right away. That part is critical. Clinical trials involve many steps from IRB review to ensure safety, back-and-forth with industry sponsors, contracting, and budgeting. All of that takes significant time and planning. Meanwhile, companies often have tight timelines and limited budgets, especially in rare diseases. What worries me most is seeing potentially life-changing therapies delayed simply because we could not move fast enough through those processes or start recruitment early enough.

That challenge is even greater in rare and ultra-rare conditions. Inclusion and exclusion criteria work well in common diseases, but when you only have five or ten potential patients, those criteria can quickly narrow the pool to just a few. Not every family is ready or able to participate in a trial, so engaging and supporting those families becomes incredibly important. I am encouraged that recent policies are starting to recognize these realities and make it easier to run trials in very small populations.

Beyond trials, widespread adoption is another major gap. Even once a therapy is approved, there is often a long delay before it reaches the patients who could benefit from it. That is especially frustrating in genetics, where only a small percentage of conditions currently have approved treatments. We know there are patients out there who could benefit, but they may not yet be diagnosed or connected to the right care team. But this is where technology can make a real difference. Advances in genetic testing, combined with tools like AI, can help flag medical records where a patient may benefit from a genetics evaluation or additional testing. That kind of early identification could lead to better outcomes and faster access to care.

At the same time, delivering these new innovations comes with real challenges, especially in the case of gene therapies. These treatments are often very high cost, and payer models are still evolving. Hospitals, payers, families, and policymakers all need to be aligned, and that takes a lot of coordination and advocacy. These therapies are here now, and patients need them, so figuring out sustainable models to deliver them is essential.

Finally, I am particularly excited about the number of therapies for ultra-rare conditions. These are diseases that may never have enough patients to support traditional drug development, yet the technology now exists to create individualized treatments. Seeing policy and regulatory pathways begin to emerge for these approaches is incredibly encouraging. It gives families hope that even the

rarest conditions may one day have a treatment within their lifetime.

How willing and prepared are patients and families to participate in clinical trials, and what types of support or resources are most important in helping them understand and navigate the decision to take part in research?

The first and most important piece is trust. especially for families dealing with very challenging and burdensome conditions. When we think about clinical trials, we have to recognize that families are already carrying the weight of the disease itself. On top of that, trials often require frequent visits, procedures, and interventions. That adds another layer of burden, and we need to be very honest and thoughtful about it from the start.

In pediatrics, families want what is best for their child, but participation also means time away from work, coordinating care for other children, and managing daily life. The human side of this is just as important as the science when it comes to trial participation. We have to acknowledge those realities and actively look for ways to support families where we know the gaps exist.

For some families, the scientific aspect is very motivating. They want to see the data, understand the mechanism, and follow the research closely. For others, it is much more about the potential outcome and whether this could help their child in the future. At the same time, there is a lot of uncertainty. That is the nature of clinical trials. Families have to grapple with things like placebo-controlled designs, which can be especially difficult to accept when you are hoping for a direct benefit. Because of these things, their decision to participate in a trial cannot be rushed. They require multiple conversations, clear explanations, and ongoing support. To be a successful trial site, we have to meet families where they are. One of the most effective drivers of participation is word of mouth. The experience a family has during a trial often determines whether other families choose to participate. That is why building strong relationships with patient and family support groups even before a trial start is so important.

You cannot run a meaningful clinical trial without first understanding the condition. That means building registries, studying natural history, and learning what daily life actually looks like for these families. This is why patient-reported outcomes are also critical. They often capture aspects of the disease that clinical measures miss and can provide a very different perspective on what truly matters to patients and caregivers. We still struggle to fully integrate these outcomes into routine care after a drug is approved, largely because of time constraints and system limitations. But as technology evolves, and with the help of AI tools, I am hopeful that tracking these outcomes will become easier and more routine.

What message would you like to share with the broader rare disease community about what meaningful collective progress looks like in advancing care and outcomes?

We are probably living through the most exciting moment in the history of genetic medicine. Over the course of my career, we have gone from diagnostic yields of around 5 percent to 40 or 50 percent, and that number will continue to rise as technologies improve. On the treatment side, we have moved from only 1 to 2 percent of genetic conditions having available therapies to closer to 5 percent today. I truly believe we are at an inflection point where that number could increase very quickly.

But progress does not happen automatically. As a system, we need to be aligned so patients can actually benefit from these advances. It is not just about having better diagnostics or new therapies, itâ??s also about figuring out how to implement them effectively. That means rethinking payer models, how care is delivered in hospitals, and how different parts of the healthcare system work together.

Many of the children we see hospitalized today likely have underlying genetic conditions, and earlier intervention could significantly change their trajectory. So if we do this right, success may actually look like fewer children in the hospital. That would be very positive because it would mean patients are being diagnosed earlier, treated sooner, managed more effectively, and live healthier and fuller lives.

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