Danielle Carnival – CEO, Undiagnosed Diseases Network Foundation (UDNF)

Many systems are built to support people once they have a diagnosis. UDNF works one step earlier, alongside individuals and families living without answers

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At a moment when rare disease policy, diagnostics, and patient-driven research are converging, Danielle Carnival sets out a clear vision for the role of the Undiagnosed Diseases Network Foundation. Drawing on her experience across neuroscience, federal health policy, and patient-led organisations, she explains how UDNF operates upstream of diagnosis, addressing the gaps faced by individuals and families living without answers. The discussion explores how data, policy, and community can be better aligned to shorten the diagnostic journey and reshape how systems respond to uncertainty.

What shaped your career path, and what led you to take on the CEO role at the Undiagnosed Diseases Network Foundation?

I have a PhD in neuroscience from Georgetown University, where my training focused on understanding how the brain functions at the molecular and cellular level. That scientific grounding shaped how I think about disease, evidence, and systems. Rather than pursuing a traditional academic path, I moved into science policy and nonprofit leadership, with a consistent focus on translating scientific insight into tangible outcomes for patients and communities.

I joined the White House around 2010, initially working on science policy priorities, including STEM education and efforts to increase diversity in the scientific workforce. Over time, my work shifted

more decisively toward health and biomedical policy, particularly in cancer and patient outcomes. I led the White House Cancer Moonshot, first under then Vice President Joe Biden and later under President Biden's relaunched initiative. Between White House roles, I served as vice president of the Biden Cancer Initiative and later became the inaugural CEO of I AM ALS, a patient-driven organisation focused on amyotrophic lateral sclerosis.

What ultimately drew me to the Undiagnosed Diseases Network Foundation was the mission. Many systems are built to support people once they have a diagnosis. UDNF works one step earlier, alongside individuals and families living without answers. The idea of building a community around that shared uncertainty, and of making practical, meaningful improvements for people facing complex and unresolved challenges, strongly resonated with me.

How do you describe the mission of the Undiagnosed Diseases Network Foundation, and how does it complement the work of the Undiagnosed Diseases Network?

The Undiagnosed Diseases Network originated as the National Institutes of Health Undiagnosed Diseases Program in 2008 and was expanded into a national research network in 2013-2014 through the NIH Common Fund. It brings together clinical sites, research cores, and a coordinating centre to tackle some of the most difficult diagnostic cases. Over the past decade, the network has evaluated more than 3,000 individuals, provided diagnoses to roughly 1,000 patients, and identified 91 previously uncharacterised conditions, contributing both to rare disease diagnosis and to broader biological understanding.

The Undiagnosed Diseases Network Foundation was created by patients and caregivers who participated in the network and recognised the need for a complementary organisation focused on the lived experience of being undiagnosed. UDNF exists to support and extend the work of the network by addressing needs that sit beyond the scope of a research programme alone.

Our work is organised around four focus areas. Support includes patient navigation and practical resources for individuals living undiagnosed. Community centres on creating meaningful connection for people who often lack a natural peer group. Action covers advocacy, policy, and reimbursement issues affecting rare and undiagnosed populations. Innovation focuses on patient-driven research questions and on strengthening the bridge from being undiagnosed to receiving a diagnosis and, ultimately, accessing treatment, ensuring people are supported throughout that journey.

How does the diagnostic journey typically unfold for people living with rare or undiagnosed conditions in the United States, and where does UDNF engage along that path?

We define our community broadly as everyone involved in the journey toward a diagnosis, from patients and caregivers to clinicians, geneticists, nurses, and researchers. Our close affiliation with the Undiagnosed Diseases Network gives us direct access to professionals who focus specifically on complex diagnostic cases, and we are building on that by expanding partnerships with additional institutions and care providers. We also see diagnostic companies as an important part of this ecosystem, given the pace of innovation in diagnostics, and we want to ensure that patient and caregiver perspectives are clearly reflected in how those tools are developed and prioritised. In that sense, UDNF sits naturally at the intersection of clinical care, research, industry, and lived experience, helping to ease the path to diagnosis.

What we see in practice is a spectrum of challenges. For some individuals, particularly those with rare or ultra-rare conditions, the science itself is still catching up, and comprehensive approaches such as whole genome sequencing are required to understand what is happening at a genetic level. In these cases, the Undiagnosed Diseases Network has demonstrated strong effectiveness, and continued scientific innovation remains essential to expanding diagnostic possibilities and, over time, treatment options.

At the same time, many people remain undiagnosed not because the science is unavailable, but because the healthcare system is difficult to navigate, fragmented, or costly. Patients with complex, multi-system symptoms often do not fit neatly into a single specialty, which the system is not well designed to handle. Through our support and community work, we are focused on providing navigation, shared resources, and peer connections that help people learn from prior experience, ask the right questions, and address barriers such as reimbursement, so that when a diagnosis is possible, there is a clearer and more supported route to reach it.

How does the undiagnosed population differ across age groups, and how do those differences influence diagnostic approaches and clinical practice?

Our experience so far has largely been shaped by the Undiagnosed Diseases Network, so that is the population I can speak to most directly. The network works with both paediatric and adult

patients, with slightly more than half on the paediatric side. In children, we most often see rare and ultra-rare conditions, particularly neurodevelopmental disorders, which continue to drive a significant proportion of diagnostic activity.

In the adult population, the picture is different but equally complex. Many individuals present with neurological or seemingly neurodegenerative symptoms, often alongside immune or autoimmune features, that do not align neatly with a single clinical specialty. These multi-system presentations can appear disconnected until an underlying cause is identified, which makes the diagnostic process more challenging and often prolongs the search for answers.

At a system level, there is no single national standard for genetic or ultra-rare disease diagnostics beyond established newborn screening programmes, which themselves continue to evolve as scientific knowledge advances. While the cost and availability of genetic sequencing have changed dramatically over the past decade, making comprehensive genomic analysis far more accessible, there is still no consistent framework for how and when it should be applied. Working alongside the Undiagnosed Diseases Network, we see a strong opportunity to better define what it means to be undiagnosed, identify those patients earlier within healthcare records, and demonstrate which diagnostic approaches deliver the most benefit for specific populations. That is where we believe meaningful progress can be made.

What policy and system-level changes would most meaningfully improve diagnosis for people living with rare and undiagnosed conditions?

There is a clear opportunity right now in rare and ultra-rare disease, particularly because many individuals who are currently undiagnosed will ultimately fall into those categories once an answer is reached. From a research funding perspective, there is growing alignment across Congress and the current administration that sustained investment in the Undiagnosed Diseases Network is beneficial. An estimated 30 million people in the United States are living with rare and ultra-rare diseases, and the experience of the Undiagnosed Diseases Network has shown that research in rare and ultra-rare conditions does not sit in isolation, but contributes directly to broader advances in our understanding of disease biology.

Regulation is another area where change could have a meaningful impact, particularly through earlier and more structured engagement with the Food and Drug Administration’s rare disease programmes. There is value in thinking about regulatory approaches and clinical trial pathways before a definitive diagnosis is reached, and in ensuring that the perspectives of undiagnosed and

rare disease communities are represented in those conversations. Bringing the patient voice into regulatory discussions at an earlier stage can help shape frameworks that are better aligned with the realities of complex and evolving conditions.

Reimbursement remains one of the most persistent structural barriers. In the United States, access to care is closely tied to ICD-10 diagnosis codes, yet many rare and undiagnosed conditions lack clear or specific coding pathways within electronic health records. This makes it more difficult for patients to secure coverage for diagnostic testing and ongoing care, as payers typically require a defined diagnosis to justify reimbursement. While symptom-based or provisional diagnoses can sometimes unlock access, they may also slow progress toward a definitive answer. Addressing this challenge will require coordinated work across patient communities, policymakers, and payers to create pathways that support access while allowing the diagnostic process to continue.

How is the Undiagnosed Diseases Network Foundation approaching collaboration across the broader rare disease ecosystem?

Although I am still relatively early in my role, collaboration is already a clear priority for us. UDNF has engaged with organisations such as the EveryLife Foundation for Rare Diseases through community and policy-related work, and we place strong value on working alongside the broader rare disease community, including organisations like the National Organization for Rare Disorders. These relationships are important for aligning efforts around patient support, research, and advocacy, and for ensuring that the perspectives of people living with rare and undiagnosed conditions are consistently represented across policy and system-level conversations.

Beyond advocacy, we are also closely watching the momentum around therapeutic discovery in rare and ultra-rare disease. One of the strengths of the Undiagnosed Diseases Network is that participants are highly data-rich, which creates an opportunity for UDNF to serve as a bridge between patients, researchers, and emerging discovery initiatives, including work focused on new models for therapy development and drug repurposing. As we grow, our role will increasingly be about connecting these efforts in a more deliberate way to help accelerate progress for people who currently have no clear treatment path. While we are still early in that process, many of these collaborations build on longstanding relationships within the rare disease community, allowing us to move forward with continuity and purpose.

What priorities are shaping your mandate at the Undiagnosed Diseases Network Foundation as you look ahead?

What initially drew me to UDNF was the strong foundation already established through the Undiagnosed Diseases Network and the depth of impact that work has had. Looking forward, I see UDNF as a place where people living with rare, ultra-rare, and undiagnosed conditions can come not only to access support, resources, and shared knowledge, but to take an active role in shaping what comes next. A central priority for us is ensuring that the community is genuinely involved in defining where we focus our efforts and in identifying the questions that still need answers.

My background in oncology and through the Cancer Moonshot has shown how much progress accelerates when patients are positioned as drivers of research rather than passive participants. Many cancers are rare, and meaningful advances came when patient input helped guide priorities and data was shared through coordinated platforms. As the broader rare disease community increasingly moves in this direction, it becomes essential for organisations like ours, which operate across multiple conditions, to stay closely connected. By working together and building shared infrastructure, we can avoid unnecessary duplication and make it easier for patients to drive research efficiently, without having to rebuild systems each time they seek answers.

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