STRATEGIC PLAN

2023-2025

Undiagnosed Diseases Network Foundation

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DEAR FRIENDS AND SUPPORTERS:

It is with great enthusiasm and a deep sense of purpose that I introduce to you our Strategic Plan for the years 2023 to 2025.

Our mission is clear: to improve access to diagnosis, research, and care for all individuals with undiagnosed and ultra-rare conditions. The UDNF has emerged as a beacon of hope and support for patients, families, clinicians, and researchers navigating the challenging landscape of undiagnosed and ultra-rare diseases.

Our strategic plan is built on the foundation of compassion, empathy, and respect, values that guide every decision we make. As we embark on this journey, we are committed to addressing the unmet needs of our community and creating an environment where no patient or family faces the unknown alone.

As we move forward, social equity will be a core value that underpins all our efforts. We are dedicated to ensuring that historically marginalized communities have equal access to diagnosis, research, care, and therapeutics. Together, we will work tirelessly to combat structural inequities and promote fairness and inclusivity both for and within the rare disease community.

I extend my heartfelt gratitude to the diverse group of stakeholders who have contributed to the development of this strategic plan. Patients, families, scientists, clinicians, and partners have all played a pivotal role in shaping our direction, and I am inspired by their dedication and passion.

In closing, I want to emphasize that this strategic plan is a living document, one that will evolve with the changing needs of our community. Together, we will create a brighter future for undiagnosed and ultra-rare disease patients, where they are no longer alone in their journey. With your support, the UDNF will be a source of strength, knowledge, and hope for all those who seek answers.

Thank you for your unwavering commitment and support.

Sincerely,

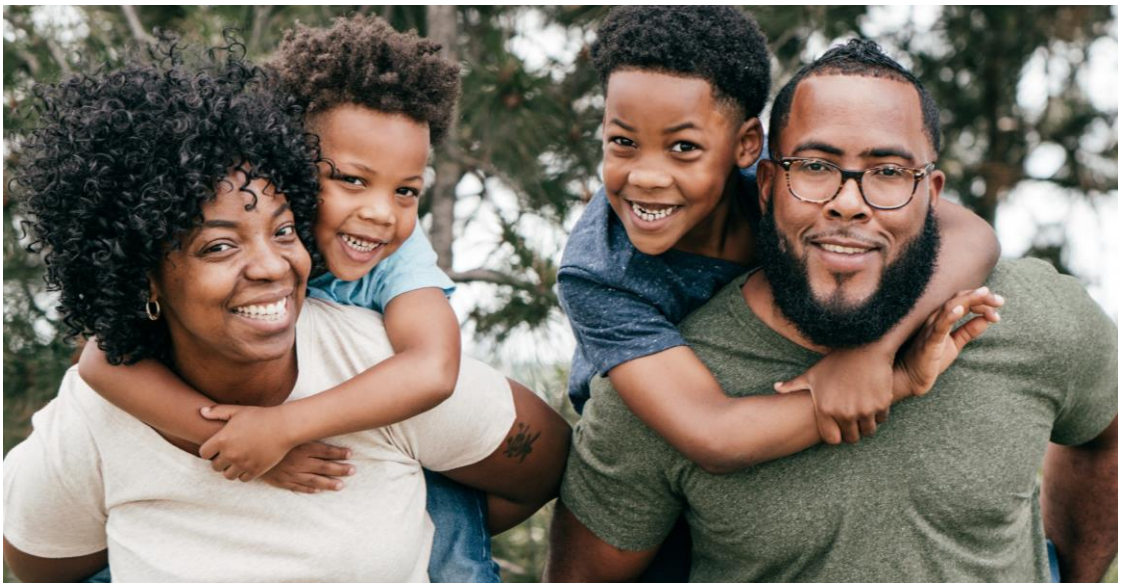
Meghan Halley, Ph.D.

President, Board of Directors
Undiagnosed Diseases Network Foundation

WHO WE ARE

“The UDN is pioneering a new personalized medicine model for helping those patients who have historically been the most difficult for the medical community to diagnose. By bringing together a nationwide network of top clinicians and laboratory researchers using the most up-to-date medical technology and knowledge, the UDN is able to provide hope for these patients, and in many cases, discover a diagnosis.”

JAMES M. ANDERSON, M.D, PH.D
Director of the National Institute of Health’s Division of Program Coordination, Planning, and Strategic Initiatives



OUR MISSION

Our mission is to improve access to diagnosis, research, and care for all with undiagnosed and ultra-rare conditions.

OUR VISION

We envision healthcare that embraces the unknown and pursues clinical and research solutions for patient well-being.

WHO WE SERVE

We provide services to undiagnosed and ultra-rare patients of all ages and their family members.

WHERE WE OPERATE

We partner with the Undiagnosed Diseases Network (UDN) sites across the country. https://undiagnosed.hms.harvard.edu/udn-sites/

OUR VALUES

"The UDN has promised to never 'tap out' and never give up. For this, we are eternally grateful. We don't know what the future has in store for Blake, but we take great comfort in the knowledge that, not only is the UDN working to find a diagnosis/cure/treatment for Blake, but that Blake will provide answers for children with rare neuromuscular conditions."

CHAD SMITH (AKA "CHAD THE DAD")
Parent of a UDN Participant

OUR CORE VALUES

COMPASSION, EMPATHY, & RESPECT
We treat patients, families, staff, board members, volunteers, and partners with kindness, understanding, and fairness. We are committed to creating a welcoming and inclusive environment.

SOCIAL EQUITY
Historically marginalized communities experience greater obstacles to accessing diagnosis, research, care, and therapeutics. We are committed to ensuring that all patients have equitable access to healthcare.

PATIENT & FAMILY-CENTERED
We believe a patient and family-centered organization can advocate for the highest quality of care and support.

INTEGRITY
We are committed to meeting the highest standards for transparency and accountability in all aspects of our operations. We go above and beyond legal requirements to share the “why” and “how” of what we do.

INNOVATION, DISCOVERY, & SCALABILITY
We invest in research and development, education, and training to foster a culture of curiosity and exploration. We create systems and processes that are efficient, effective, and sustainable.

PARTNERSHIPS & COLLABORATION
By working with a diverse cross-section of stakeholders, we are committed to expanding best practices for discovering diagnoses and therapies for undiagnosed and ultra-rare disease patients.

OUR HISTORY

“It was nothing short of amazing. Our call ended with my full understanding that these were not just findings in some abstract research activity, not just some manuscript being submitted for the benefit of the annals of science. This was the end of my daughter’s diagnostic odyssey. This was a diagnosis. Now the odyssey for a cure can begin.”

SARAH MARSHALL
UDN PEER Co-Chair
Parent of a UDN Participant

2023

Patient advocates secured $18M through congressional budget appropriations to support ”Phase 3” of the UDN. This new phase marked the start of a new model led by and for patients.

2022

Members of the UDN PEER and UDNI founded the UDNF in response to dissipating funds from the NIH Common Fund.

2018

The NIH launched “Phase 2” of the UDN by expanding from seven to twelve clinical sites and establishing an additional model organisms screening center.

2013

Building on the success of the UDP, the NIH launched the Undiagnosed Diseases Network (UDN) through the Common Fund. ”Phase 1” included six additional clinical sites and multiple research cores.

2008

The Undiagnosed Diseases Program (UDP) was established at the NIH to uncover, understand, and treat rare disease.

NEEDS ASSESSMENT

“The unsettling and stressful feelings I previously had are gone. They are not gone because I’m cured or on a therapy plan. Or even received a diagnosis yet. No, they are gone because for the first time in this journey, I truly believe the best team is working on my case. Instead of being limited to matching my symptoms to a well-known disease, which obviously hasn’t worked, the UDN is working beyond those limits. The calming feeling the UDN has brought to my life, to me, is the best example I can give of the value the UDN provides, not only to those for whom a diagnosis has been made, but to all UDN participants.”

TROY EVANS
UDN Participant
Board Treasurer, UDNF

UNDIAGNOSED DISEASES NETWORK

Research studies funded by the NIH Common Fund are limited to 10 years of support, meaning that Common Fund support for the UDN ended in June 2023. Thanks to the advocacy of undiagnosed patients and families, the Consolidated Appropriations Act of 2023 provides financial support for the Coordinating Center, clinical sites, and research cores through September 2023, and more limited support for the Coordinating Center and clinical sites from 2023-2028.

PATIENT AND FAMILY NAVIGATION

Diagnosis is just one of the many unmet needs faced by patients and families with undiagnosed and ultra-rare diseases. Patients report frustrations with the limited communication, lack of coordination, and unclear expectations regarding processes, timelines, and post-diagnostic options. Even when a patient receives a diagnosis, after this often-confusing evaluation process, they likely will not have an available therapeutic option.

SYSTEMIC INEQUALITY

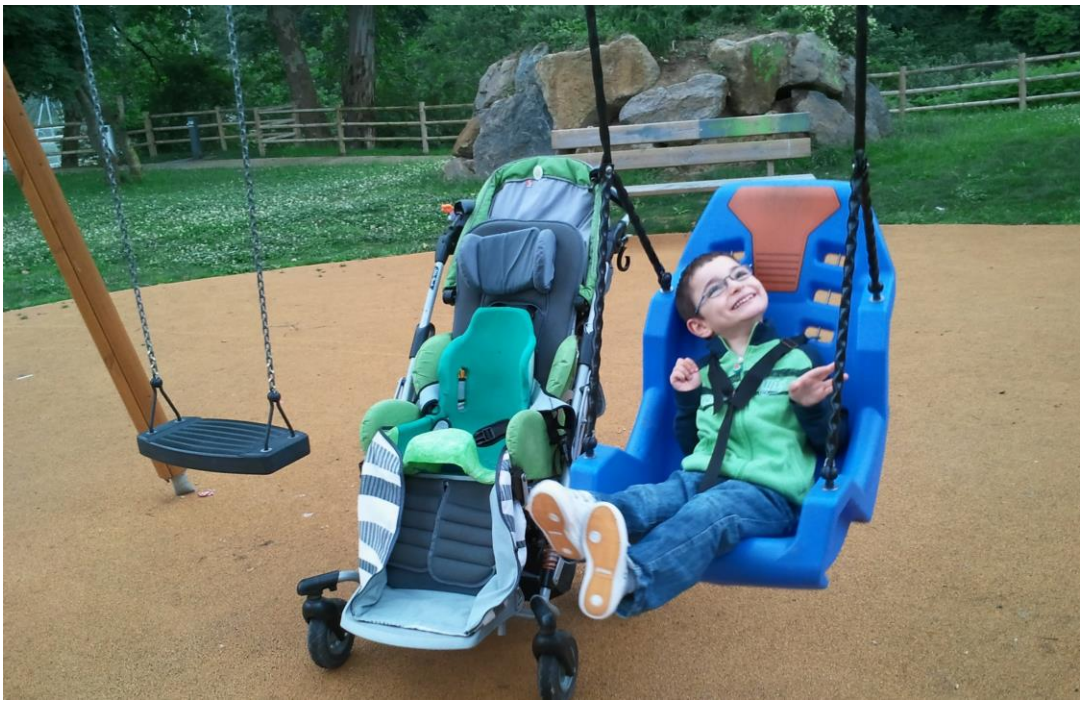
The needs assessment identified limited resources and support for outreach to underserved communities. Despite a commitment to accessibility and funds available to cover travel and other expenses for patient participants and their families, White families remain overrepresented among network participants.

Considering that 30 million individuals live with a rare disease in the U.S. and a rare disease diagnosis takes an average of 7 years, millions of individuals currently face the uncertainty of an undiagnosed disease.

THE NEXT THREE YEARS

“They continue to research her genes and her conditions to find a diagnosis. We will never give up hope. We hope that someday we can find a word or phrase that could help describe Kaci to others. We hope that it will be something you can look up on a computer and better understand. We hope that we have a community of others with the same diagnosis so that we can support each other. We hope to be able to help and mentor the new parents with a recently diagnosed child. We hope for someone with a child older than Kaci, to tell us what our future with her might look like.”

TAMMI CREED
Parent of a UDN Participant



Since 2013, the UDN has set the international standard for researching undiagnosed diseases. In just under a decade, the UDN reviewed 5,882 applications, accepted 2,354 participants, evaluated 1,946 patients, and diagnosed 546 children and adults. Because the NIH Common Fund grant that established the UDN was set to end in June 2023, undiagnosed patients and families across the country, along with researchers and clinicians, committed to coming up with new strategies to sustain and expand the UDN.

Undiagnosed patients, families, clinicians, and researchers conducted a needs assessment during the summer of 2022 to better understand what this change could mean. The team identified several opportunities, in addition to continuing the UDN, to improve outcomes for patients. “I want to see the UDN sites not only continue but to [also] grow and make even more discoveries and [...] impacts on the lives of patients and families,” explained Michele Herndon, mother of a UDN participant.

OUR PRIORITIES

The UDNF will fill a unique need for ultra-rare and undiagnosed disease patients who currently have to navigate numerous unrelated services and supports. The UDNF will be at the center of research, therapeutics, clinical care, and diagnosis to provide comprehensive services to undiagnosed and ultra-rare patients and their families under one roof. We will foster partnerships between patients and families, advocacy organizations, researchers, payers, industry, funders, venture philanthropists and healthcare providers to advance individual and community well-being.

1. ORGANIZATIONAL GROWTH & SUSTAINABILITY

The UDNF's growth and sustainability efforts will ensure that it is a sustainable organization for the long term. We will focus on three key areas: fundraising, development, and financial sustainability.

ACTIVITIES

- Create and implement an annual sustainability and growth plan for long-term funding starting in 2023.
- Integrate social and economic equity values throughout the organization's governance, policies, procedures, budgets, and activities.
- Adopt a governing board model in 2023 that tasks the CEO with carrying out the strategic plan set by the board of directors.
- Partner with aligned organizations to maximize the UDNF's mission.
- Develop a staffing plan for the UDNF that supports the strategic priorities and activities.

On average, a rare disease patient will see 12 specialists before receiving a diagnosis.

2. COMMUNITY ENGAGEMENT & OUTREACH

The UDNF is committed to building a community by and for all patients with undiagnosed and ultra-rare diseases and their families. We will serve as the leading resource for trustworthy information about these diseases and services, and we will ensure that underrepresented patients have equitable access to diagnosis, research, therapeutics, and support.

ACTIVITIES

- Develop and implement an annual communications plan.
- Partner with existing organizations to add and enhance undiagnosed resources and build in-person communities.
- Develop a community engagement and outreach strategy with key programs and routinely review and revise programs and procedures to increase accessibility and inclusivity for diverse communities.
- Engage undiagnosed patients and families online through virtual events, groups, message boards, and posts.
- Educate frontline medical and social service providers, including genetic counselors, about undiagnosed and ultra-rare diseases, especially those most likely to work with underserved patients.

Historically marginalized communities may experience lower diagnosis rates for rare diseases due to the inequitable distribution of resources and living in areas with few practicing specialists.

3. PATIENT & FAMILY NAVIGATION

We are committed to improving medical management, care coordination, access to social services, support, and mental health services for patients with undiagnosed and ultra-rare diseases and their families. We will also improve the patient and family experience of the diagnostic process and transition to therapeutics, regardless of when or whether a patient receives a diagnosis. We will use consolidated tools and strategies across undiagnosed and ultra-rare disease communities.

ACTIVITIES

- Design and pilot a patient and family navigation program for UDN participants, including a thorough evaluation process for program impact.
- Develop a living resource database on the website for undiagnosed and ultra-rare patients, UDN participants, and non-UDN participants with information in multiple languages about social services, health coverage, medical providers, financial support, diagnostic options, and local and national resources.
- Support partnerships between patients, families, clinicians, and scientists to drive diagnosis, care, and therapeutic matching for ultra-rare, newly diagnosed, partially diagnosed, and undiagnosed disease patients.

To date, the UDN has received 6,570 applications, accepted 2612 patients, evaluated 2,220 patients, and diagnosed 676 patients.

4. PATIENT-CENTERED RESEARCH & CLINICAL CARE

The UDNF is committed to making a diagnosis of ultra-rare diseases more accessible, equitable, accurate, and efficient. We will do this by advancing the diagnosis and knowledge of these diseases through patient-centered research.

ACTIVITIES

- Improve patient outcomes by collaborating with the UDN and other key stakeholders.
- Support efforts to drive therapeutic matching for ultra-rare and newly diagnosed diseases.
- Secure and provide financial support to the UDN.
- Identify and build a network of non-profit, industry, and academic partners committed to a shared ethos of patient-centered care for undiagnosed and ultra-rare diseases.
- Support efforts to streamline data collection, increase access, and enhance entry for UDN participants and applicants.
- Develop a strategy and support efforts to drive an innovative model for diagnosis of participants within the UDN.
- Broaden the expertise and types of specialists involved with the UDN, including for non-genetic disorders.

Approximately 350 million people have a rare disease. About 80% of rare diseases are genetic, and only 5 percent have an FDA approved treatment.

THANK YOU

“Bewildered, I reached out to our nurse coordinator at the UDN who assured me that the search for a diagnosis does not end with her death. The UDN is still committed to finding a reason why my sweet and beautiful 21-month-old girl died. They are invested in our family and helping to give us some bit of peace. Not knowing why your child died is torture. The UDN gives me hope that one day, we will know what took her from us. It also gives me hope that once identified, it will provide valuable information for my children as they grow and start their own families.”

NIKKI PATRICK
Parent of a UDN Participant

THANK YOU

We developed this strategic plan through the hard work and dedication of a diverse group of stakeholders from the undiagnosed, rare, and ultra-rare disease community. We are grateful for the contributions of everyone who participated in the planning process, including our board of directors, staff, volunteers, patients and families, scientists and clinicians, and partners. We believe this plan will help us achieve our mission and vision for the future. It is a living document that will be updated to remain relevant and effective.

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UDN team members
Patients and families
Partners
Personnel and consultants

UNTIL RARE IS NOT A BARRIER

Undiagnosed Diseases Network Foundation