2023 annual report

Undiagnosed Diseases Network Foundation

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Mission & Vision

At the Undiagnosed Diseases Network Foundation, our mission is clear: to improve access to diagnosis, research and care for all individuals with undiagnosed and ultra-rare conditions. We are dedicated to improving diagnostic strategies and treatments, providing answers where there were none before.

Our vision is equally compelling: healthcare that embraces the unknown and pursues clinical and research solutions for patient wellbeing. We believe that through collaboration with healthcare professionals, researchers, and advocates, we can achieve this vision and bring hope to countless individuals and families.

In 2023, our mission and vision guided every aspect of our work, from launching our patient navigation and support services to driving forward advocacy efforts to ensure funding for the Undiagnosed Diseases Network. As we look to the future, these principles will remain at the heart of all that we do.

Troy Evans, UDNF Treasurer

Welcome note

Dear Friends and Supporters,

As we reflect on the launch of the Undiagnosed Diseases Network Foundation (UDNF), we are filled with gratitude for the unwavering support that has fueled our growth this past year. Our journey began in response to the pressing need for sustainability of the Undiagnosed Diseases Network (UDN) and to address the gaps in support for the undiagnosed and ultra-rare disease community.

The UDN's legacy has provided a strong foundation for our work, and we are committed to building upon its achievements. Through the establishment of UDNF, we aim to create a more sustainable and equitable future for individuals affected by these conditions.

One of our most significant milestones this year has been the successful launch of our Patient Navigation Program. This initiative is made possible by a generous $2.5 million grant from the Chan Zuckerberg Initiative (CZI), which has provided us with the resources necessary to build the UDNF's infrastructure and provide critical support to patients and families navigating the diagnostic and therapeutic odyssey.

We are also deeply grateful for the dedication and expertise of our inaugural Board of Directors. Their guidance and support have been invaluable in shaping our organization's first strategic plan which will guide the direction of our organization from 2023 through 2025, ensuring that our mission and four strategic pillars remain at the forefront of our efforts.

As we continue to navigate the challenges and opportunities that lie ahead, we are confident in our ability to make a lasting impact on the lives of those affected by undiagnosed and ultra-rare diseases. Your continued support is essential to our mission, and we are grateful for your partnership in this important endeavor.

As you read through this report, we hope you feel the same sense of hope and determination that drives our work every day. Together, we are building a community for those who need it most. Thank you for your continued support.

Sincerely,
Meghan Halley
Amy Gray

Meghan Halley
Board Chair, UDNF

Amy Gray
Chief Executive Officer, UDNF

Strategic Plan

To guide our efforts, we have developed a comprehensive strategic plan with four key pillars:

SUSTAINABILITY & GROWTH
Ensuring the long-term viability of the UDN network and our organization, expanding our reach to serve more patients.

PATIENT & FAMILY NAVIGATION
Providing personalized support and guidance to individuals navigating the complex healthcare system.

COMMUNITY OUTREACH & ENGAGEMENT
Building strong partnerships and fostering a sense of community among patients, caregivers, and healthcare providers.

PATIENT-CENTERED RESEARCH AND CLINICAL CARE
Advancing research and improving clinical care to enhance the lives of individuals with undiagnosed and ultra-rare diseases.

This strategic plan is more than just a roadmap; it is a commitment to our mission and to the patients and families we serve. We are excited about the future and confident that this plan will guide us toward achieving our goals.

Grant

IN 2023, UDNF WAS HONORED TO RECEIVE A TRANSFORMATIVE $2.5 MILLION GRANT FROM THE CHAN ZUCKERBERG INITIATIVE (CZI). THIS GRANT HAS BEEN A CORNERSTONE OF OUR EFFORTS TO PROVIDE SUPPORT TO PATIENTS AND FAMILIES, ENHANCE RESEARCH, AND ADVOCATE FOR THOSE WITH UNDIAGNOSED AND ULTRA-RARE DISEASES.

THE IMPACT OF THE CZI GRANT HAS BEEN PROFOUND. WITH THESE FUNDS, WE HAVE HIRED A TEAM OF PATIENT NAVIGATORS, LAUNCHED THE FIRST UDNF-FUNDED RESEARCH PROJECT, HELD PATIENT AND FAMILY EDUCATION MEETINGS, PROVIDED EDUCATIONAL PROGRAMMING TO HEALTH-CARE PROFESSIONALS, AND HOSTED OUR FIRST ANNUAL UNDIAGNOSED DAY ACTIVITIES. WE ALSO EXPANDED OUR PATIENT SUPPORT SERVICES, DEVELOPING NEW RESOURCES AND SUPPORT MATERIALS FOR INDIVIDUALS NAVIGATING THE COMPLEXITIES OF THE HEALTHCARE SYSTEM.

LOOKING AHEAD, THE CZI GRANT WILL CONTINUE TO EMPOWER OUR WORK, ENABLING US TO PUSH THE BOUNDARIES OF WHAT IS POSSIBLE IN THE FIELD OF UNDIAGNOSED AND ULTRA-RARE DISEASES. WE ARE DEEPLY GRATEFUL FOR THIS PARTNERSHIP AND THE OPPORTUNITIES IT HAS CREATED.

- **6 Navigators Hired**
- **Intro to Patient Navigator Program**
  - 78 Attendees - Together on Tuesdays

PEER

The Undiagnosed Diseases Network Foundation (UDNF) has undergone a significant transformation, merging with the Patient Engagement and Empowerment Resource (PEER) to create a more inclusive support network for individuals and families affected by undiagnosed and ultra-rare diseases. This integration enriches the foundation's mission by uniting resources and initiatives that reflect the voices and needs of the community. One of the key goals for 2024 is to maintain the quarterly PEER newsletter, a vital communication tool that keeps the community informed on advancements, events, and resources tailored to support patients and caregivers. Additionally, the Tell Me More Series will remain a cornerstone of UDNF's efforts to share personal stories and insights from individuals on their diagnostic journeys. These programs highlight UDNF's commitment to fostering a space where patients, caregivers, and advocates can connect, learn, and find encouragement as they navigate complex healthcare challenges.

The integration of UDNF and UDN PEER marks a pivotal step in amplifying the voices of patients and families within the undiagnosed and ultra-rare disease communities. UDN PEER provides critical support by sharing resources, collaborating with researchers, and raising public awareness. This partnership will empower those affected by undiagnosed diseases with a stronger voice and the tools needed to navigate their journeys. Together, UDNF and UDN PEER will strengthen the community, offering patients and families hope for a diagnosis and research that could pave the way for treatments.

Building Our Founding Team

In 2023, we were thrilled to assemble our inaugural team at the Undiagnosed Diseases Network Foundation (UDNF). As a newly established organization, this was a critical step in laying the groundwork for our future success.

Leadership and Strategy

Amy Gray, who also serves as Co-Chair of the UDN Executive and Steering Committees, joined us as our Inaugural CEO. Amy's expertise and leadership have been invaluable in developing our strategic plan and shaping the direction of UDNF.

Patient Navigation and Support

Michele Herndon joined our team as Program Director, bringing her passion for patient advocacy and deep understanding of the healthcare system. Under Michele's leadership, we successfully launched our Patient Navigation Program, a vital service that connects patients with the resources and support they need to navigate their complex medical journeys.

Our team of patient navigators, some with children who have gone through the UDN, has made a profound impact on the lives of countless individuals by providing personalized guidance, connecting them with specialists, and advocating for their needs. Their dedication and compassion are at the heart of our mission.

Discover more about our dedicated team and their impact by clicking **here**.

Leadership & PEER

The success of UDNF in 2023 would not have been possible without the dedication and leadership of our Board of Directors, Advisor, and Patient Engagement and Empowerment Research Group Members (PEER).

Our Board of Directors consists of leaders in healthcare, research, and advocacy who provide strategic guidance and oversight for our organization. Their expertise and commitment have been instrumental in advancing our mission. We extend our deepest gratitude to our founding board members—Meghan Halley, Troy Evans, and Helene Cederroth—whose vision, dedication, and unwavering support have laid the foundation for our success. Without their leadership and passion, we would not be where we are today. Thank you for believing in our mission and helping to shape the future of our community.

The Advisory Board offers valuable insights and advice, ensuring that UDNF remains at the forefront of research and advocacy for undiagnosed diseases. The guidance from our Board Advisor has played a vital role in shaping our programs and initiatives.

The PEER Group Members are patient advocates and experts who ensure our programs meet the needs of the undiagnosed and ultra-rare community. Their voices are essential to our work, and we are grateful for their continued support and partnership.

our board

The Undiagnosed Diseases Network Foundation extends heartfelt gratitude to Kimberly LeBlanc, Cristina Casanova Might, Adaline Dunnberg, and Jo Dunnberg whose invaluable guidance and dedication were instrumental in advising our Board and establishing UDNF. Their expertise and commitment provided a strong foundation for our mission, ensuring that UDNF is well-prepared to support patients and families in need. We thank them for their generosity of time and unwavering support in launching this organization.

MEGHAN HALLEY

Board Chair | Founding Board Member | she/her

Meghan Halley, PhD, MPH, is a Senior Research Scholar at Stanford University's Center for Biomedical Ethics, specializing in the intersection of ethics and economics in genomic technologies, with a focus on patient data sustainability, governance, and outcomes in rare disease diagnosis and pediatric care.

HELENE CEDERROTH

Outgoing Vice Chair 2023 | she/her

Helene Cederroth is a Co-founding Board Member and Outgoing Vice Chair. Helene also serves as Co-founder and Permanent Board Member of the Undiagnosed Diseases Network International (UDNI) and is the Founder and President of the Wilhelm Foundation, where she supports families and promotes research for undiagnosed diseases. Her leadership continues to shape the future of rare disease advocacy and medical advancements.

SIKA DUNYOH

Incoming Vice Chair | Founding Board Member | she/her

Sika Dunyoh, Senior Director of Patient Advocacy at Travere Therapeutics, brings extensive experience in patient advocacy, engagement, and education, inspired by her sister's battle with relapsing polychondritis; she previously led educational initiatives at the National Organization for Rare Disorders.

our board

TROY EVANS

Treasurer |Founding Board Member|he/him

Troy Evans is the Owner and President of Cinch Home Loans. He is a patient of the Undiagnosed Diseases Network (UDN) and a resident of Draper, UT. He entered the UDN as a patient in 2018 at the UCLA site. Despite not yet receiving a diagnosis, his personal experience has given him a unique perspective and deep commitment to the organization's mission and sustainability.

F. SESSIONS COLE

Secretary | he/him

Dr. Cole, a neonatologist and researcher, has over 25 years of NIH-funded experience in rare disease genomics and led the Undiagnosed Diseases Network Clinical Site at Washington University. He currently co-chairs the UDN's Sustainability Working Group and leads the Therapeutic Matching Committee, focusing on therapies for rare disease patients.

ANNE PARISER

Vice President Medical and Regulatory Affairs, Alltrna | she/her

Anne Pariser, VP of Medical and Regulatory Affairs at Alltrna, has over 20 years of experience in rare disease research, including roles at the NIH and FDA. She is an expert in advancing therapeutics for rare diseases through research, collaboration, and policy development.

EFFIE PARKS

Host, Once Upon a Gene Podcast | Rare Disease Community Influencer/Advocate/Storyteller | she/her

Effie is a dedicated advocate for those affected by rare genetic conditions, inspired by her child's diagnosis with CTNNB1 syndrome. As the host of the "Once Upon a Gene" podcast, she shares stories from the rare disease community to educate, connect, and amplify their voices.

BOARD OF DIRECTORS

our board

GERALD SWEENEY

Retired Litigation Attorney | he/him

Gerald Sweeney, a retired litigation attorney with over 40 years of experience, has served on the boards of the Cystic Fibrosis Foundation and various entertainment companies. He has also participated in the Undiagnosed Diseases Network (UDN), bringing a wealth of legal, entertainment, and advocacy expertise to any board.

JESSICA SWANSON

UDNF PEER Co-Chair | she/her

Jessica Swanson is the founder and CEO of Summit Health Services Inc., specializing in Applied Behavior Analysis (ABA) with 20 U.S. locations. A Board Certified Behavior Analyst (BCBA), she also serves on the board of QABA and leads Redpoint Consulting Inc., advising start-ups and Pathfinder Health. As co-chair of UDNF PEER, she advocates for individuals with rare diseases. Jessica is married to an active-duty officer, has two children, and lives in Huntsville, Alabama.

KELLY CERVANTES

Writer, Speaker, Advocate | she/her

Kelly Cervantes is an award-winning writer, speaker, and advocate, best known for her blog “Inchstones” and her upcoming book, *Normal Broken: The Grief Companion for When it’s Time to Heal But You’re Not Sure You Want to*. She has been recognized as one of "Chicagoans of The Year" and is a former Board Chair of CURE Epilepsy, where she also hosts the “Seizing Life” podcast.

BOARD OF DIRECTORS

our board

LILIANA FERNANDEZ

Medical Monitor, Premier Research | she/her

Liliana Fernandez is a medical professional with 20 years of research experience, including 7 years specializing in undiagnosed and rare diseases. She worked at the Stanford Center for Undiagnosed Diseases, developing a workflow to enhance Hispanic participation, and now serves as a medical monitor for gene therapy clinical trials in rare diseases.

PARVATHY KRISHNAN

Chief Executive Officer,
Krishnan Family Foundation| she/her

Parvathy Krishnan, CEO and founder of the Krishnan Family Foundation, advocates for rare diseases following her children's diagnoses with ultra-rare conditions. With a Master's in Clinical Nutrition and experience as a Nutrition Support Dietitian, she engages stakeholders globally, serves on advisory councils, and brings a unique perspective to her advocacy work from her home in North Carolina.

STEPHAN ZUCHNER

Professor of Human Genetics and Neurology, University of Miami | he/him

Stephan Zuchner, M.D., Ph.D., is a Professor at the University of Miami Miller School of Medicine, specializing in rare disease genetics. His research has identified key disease genes, and he holds leadership roles in ClinGen and several advisory boards.

BOARD OF DIRECTORS

our board

STEPHANIE TOMLINSON

UDNF PEER Co-Chair | she/her

Stephanie Tomlinson has advocated for rare disease patients for over 20 years and has served on committees for early childhood special education. Formerly the Patient Support Coordinator for MitoAction, she now hosts the podcast "Energy in Action," which highlights patient stories and expert research.

TANIA SIMONCELLI

Vice President of Science in Society, Chan Zuckerberg Initiative (CZI) | she/her

Tania Simoncelli is Vice President of Science in Society at the Chan Zuckerberg Initiative, where she launched the Rare As One Project to support patient-driven research. With over 20 years in science policy, including roles at the White House and FDA, she holds degrees from Cornell and UC Berkeley and was named a 2013 “person who mattered” by Nature and an AAAS Fellow in 2021.

TOM KELLY

Founder, UnitedHealth Group Special Needs Initiative | he/him

Tom, recently retired after 13 years as V.P. of Marketing and Innovation at UnitedHealth Group, led the creation of the Special Needs Initiative, a team of 400 focused on improving healthcare for families with children with special needs. With over 16 years of experience in rare diseases, he draws from his personal experience as a father of a child with an ultra-rare genetic disorder.

UDNF

Advisor

MAX BRONSTEIN

Senior Advisor to the Board of Directors | he/him

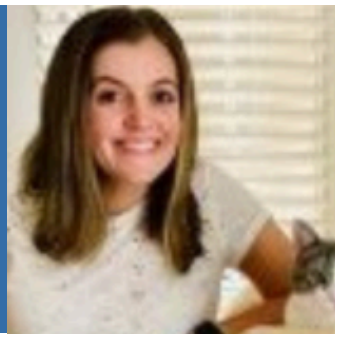
Max G. Bronstein is a mission-driven health policy entrepreneur with 18 years of experience across corporate, government, academic, and non-profit sectors. He led government affairs and patient advocacy at Crinetics Pharmaceuticals, focusing on rare endocrine diseases, and served as Assistant Director for Health Innovation at the White House Office of Science & Technology Policy, where he spearheaded initiatives like the launch of ARPA-H and efforts to improve patient access to genomic sequencing. Max also held leadership roles at Audentes Therapeutics and the EveryLife Foundation for Rare Diseases, where he helped drive the passage of the 21st Century Cures Act and advocated for improved newborn screening policies.

Max's work has been published in STAT, Pediatrics, Forbes, Nature, and the New England Journal of Medicine, and he founded the Journal of Science Policy & Governance to elevate the voices of young scholars in policy-making. He holds a master's degree in public policy from the University of Michigan and a BA in biology from Ithaca College. Outside of work, he enjoys hiking, sailing, swimming, and diving around the San Francisco Bay Area.

PEER

Kara Anderson | she/her

Kara Anderson is a postdoctoral fellow in Virginia who was diagnosed with a rare disease at the Duke site in 2018 and joined PEER that same year, finding the group invaluable in navigating her own condition. Outside of her work and advocacy, she enjoys cooking and dreams of turning her cat, Kit, into an Instagram influencer.



Sarah Marshall | she/her

Sarah Marshall, a UDN(F) PEER member since 2018, uses her social work background and personal experience caring for an undiagnosed child to advocate for those with rare diseases. She also supports women and children with chronic illnesses, focusing on immigrant and marginalized communities.

Christine McGarvey | she/her

Christine McGarvey and her daughter, Brigid, have been participants in the UDN at CHOP since 2021, both on diagnostic journeys. A rare disease advocate and NORD State Ambassador, Christine holds advanced degrees in Biochemistry and Cell and Molecular Biology and directs the UDNF PEER “Tell Me More” lecture series.

Nikki Patrick | she/her

Nikki Patrick, a mother of four from suburban Chicago, became involved in rare disease advocacy after her daughter Felicity passed away from an undiagnosed condition in 2020. She is dedicated to supporting families on similar journeys and hopes to find answers that may help others.

Casey Robertson | she/her

Casey Robertson, E.Ds., lives in Mississippi and has sought a diagnosis for her daughter Kylie, 17, who joined the UDN in 2022. She works in blindness research, advocates for underserved students and families, and edits the UDN newsletter.

Community Engagement & Outreach

In 2023, we strengthened our connection with the undiagnosed disease community by launching the Patient Navigator Program, a vital initiative offering personalized support to families navigating the diagnostic odyssey. This program became a cornerstone of UDNF's growth and impact as we prepared for 2024.

The Patient Navigator Program assists with the UDN application process, helping individuals understand the criteria, apply, and, if accepted, collaborate with their UDN site to support participation. The program also offers ongoing communication, providing timely updates and facilitating questions between UDN site staff and patients. For those not accepted into the UDN, or for individuals with ultra-rare diseases or undiagnosed conditions, the Patient Navigator Program helps guide them to resources, navigate the healthcare system, and explore other opportunities for support.

We also expanded our team in the areas of community engagement, outreach, and education. Our dedicated team members worked tirelessly to raise awareness, build partnerships, and provide valuable resources to patients and families. The assembly of our founding team was a critical step in positioning UDNF for continued success.
As we moved forward into 2024, we remained committed to advancing our mission through the four pillars that guided our work: Supporting Families, Advancing Research, Building a Collaborative Community, and Developing a Sustainable Organization for the future. We are grateful for the dedication and expertise of our team members and looked forward to the positive impact they would continue to make in the lives of those affected by undiagnosed diseases.

Financial Summary

Statement of Activities for the Year Ended December 31, 2023

We are committed to financial transparency and responsible stewardship of the funds entrusted to us. Every dollar we receive is used to advance our mission and make a difference in the lives of those we serve.

$2.58M

Total Revenue

Revenue and Support

- Grants & Contributions: $2,571,504
- Investment Income: $15,127

Total Revenue & Support: $2,586,631

$218K

Total Expenses

Expenses

- Program Services: $198,033
- Supporting Services:
- Management and General: $8,776
- Fundraising: $11,528

Total Expenses: $218,337

$2.37M

Change in Net Assets

Change in Net Assets

- Net Assets at Beginning of Year: $2,807
- Increase in Net Assets: $2,368,294

Net Assets at End of Year: $2,371,101

Year in Review

Additional Key Achievements

2023 was a year of new accomplishments for UDNF. Beyond the highlights of the CZI grant, the hiring of our team, and the development of our strategic plan, there were numerous other milestones worth celebrating.

Our advocacy efforts yielded significant results in 2023. We worked closely with policymakers, healthcare providers, and patient advocacy groups to raise awareness of the challenges faced by individuals with undiagnosed and ultra-rare diseases and to advocate for policies that support timely and accurate diagnoses. A significant victory was our successful advocacy for UDN funding for 2023, securing $18 million in funding from the National Institutes of Health to support the UDN clinical sites, research cores, and Data Management Coordinating Center. This funding will be instrumental in advancing research and improving care for patients with undiagnosed and ultra-rare diseases.

Community engagement was another area of focus in 2023. Through our outreach efforts, we were able to increase awareness of UDNF and our mission, reaching hundreds of new community members, leading to donations and volunteer support. We are grateful to everyone who has joined us in this journey and look forward to continuing to build our community in the years to come.

$2.5M

GRANT FROM THE CHAN ZUCKERBERG INITIATIVE (CZI)

$18M

CONGRESS PASSED SPENDING BILL IN 2023 TO FUND THE UDN

97

DONATIONS MADE THROUGH GIVEBUTTER.

Thank You

As we close this chapter in 2023, we extend our heartfelt gratitude to everyone who has been a part of our journey. To our donors, your generosity has fueled our mission and allowed us to make significant strides in ending the diagnostic odyssey. To our partners and collaborators, your expertise and dedication have been invaluable in advancing our shared goals. To our volunteers, your time and passion have made a meaningful impact on the lives of those we serve. And to our community, your unwavering support and advocacy continue to inspire us every day.

Stay Connected

We invite you to continue your engagement with the Undiagnosed Diseases Network Foundation. Stay up to date with our latest news, events, and initiatives by following us on social media and visiting our website.

Contact Information
Website: www.udnf.org
Email: info@udnf.org
Phone: 202-599-4465

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Instagram: @theudnf

Thank you for your continued support. Together, we can make a difference.