RDDC™

RARE DISEASE DIVERSITY COALITION

2024 RDDC ACHIEVEMENT REPORT

BLACK WOMEN'S HEALTH IMPERATIVE

RDDC

RARE DISEASE DIVERSITY COALITION

2024 RDDC ACHIEVEMENT REPORT

CONTENTS

RARE DISEASE DIVERSITY COALITION

2024 RDDC Achievement Report

The Rare Disease Diversity Coalition (RDDC) actively unites active individuals and organizations to address the unique barriers and difficulties encountered by patients from historically marginalized communities, their families, and surrounding communities.

While many might not frequently come across rare diseases, these conditions impact an estimated 25-30 million individuals across the United States.

In our efforts to address the myriad of challenges faced by patients afflicted with the approximately 10,000 known rare diseases, we must recognize and embrace the remarkable diversity within these diseases. Historically marginalized communities face significant health disparities when a rare disease impacts them. These patient populations are more likely to experience delays in diagnosis, with adverse consequences for prognosis, treatment, and quality of care. Their specific needs and experiences are less likely to be understood by healthcare practitioners. Rare diseases disproportionately affecting communities of color are less likely to attract funding for research.[1]

2024 marks the fourth anniversary of RDDC and our focus remains unwavering. We are dedicated to fostering equitable access to diagnosis and treatment for rare diseases, particularly among underrepresented populations. We significantly expanded our reach, now proudly partnering with over 90 member organizations dedicated to advancing equity in rare disease care. We are committed to amplifying their voices and addressing their unique needs, ensuring that they are not just heard, but truly understood.

RDDC firmly believes in the power of collaboration. We recognize the importance of leveraging the input of all stakeholders, including rare disease patients and families, who experience the struggles of these issues daily; patient organizations, which represent and unite those most affected; clinicians and researchers, who drive understanding and evidence across the medical community; private-sector groups, which play a crucial role in supporting research; and policymakers, who drive change through government legislation. It's through this collective effort that we can drive the changes needed to address rare diseases.

RDDC remains steadfast in its commitment to identifying and championing evidence-based solutions that address the inequitable challenges faced by individuals seeking timely diagnosis, and information and create a brighter future for those affected by rare diseases.

1 National Press Foundation. "Diversity Elusive in Rare Disease Research." Retrieved from https://nationalpress.org/topic/diversity-elusive-in-rare-disease-research/

2024 KEY ACHIEVEMENTS

In 2024, several key achievements have marked significant milestones across the Coalition including:

- **Compiled the Patient and Caregivers Barriers of Access survey results** and disseminated an Executive Summary with initial findings.
- **Successfully hosted the 2nd round of the RDDC Fellows Program** with two Fellows hosted by two patient advocacy organizations. This program aims to inspire the next generation of the medical workforce, fostering a commitment to addressing rare disease disparities, particularly among people of color, underserved, and underrepresented communications.
- **Produced and disseminated a Diversity, Equity, and Inclusion (DEI) Organizational Readiness Quiz.** This quiz aims to help leaders evaluate their organization's DEI strategy and activities to uncover areas to improve or increase their efforts.
- **Hosted the 2nd Annual "The Eve of Rare Disease Day Reception."** At the event, RDDC presented the inaugural RISE awards to a handful of rare disease health equity champions who have been Resilient, Impassioned, Strong and Empowered towards the mission and vision of the RDDC.

- **Co-sponsor of the 4th Annual Rare Health Equity Forum** scheduled for September 2024. The theme will be "Together in Equity, Driving Change". Through plenary sessions, networking, and interactive table talks, the event empowers attendees with actionable tools and strategic insights to advocate for a more inclusive rare disease community.[2]

More details about each of these and other achievements are provided throughout this report.

2 https://globalgenes.org/rare-health-equity-forum/

WORKGROUP:
Patient, Provider, Caregiver Journey

Rare disease patients and their caregivers are at the heart of RDDC's mission. To better serve these populations, we must gather data on the experiences of diverse patients with rare diseases and their caregivers. Caregivers, who often contribute selflessly and tirelessly to their loved ones' well-being, have historically been overlooked and inadequately supported. RDDC is committed to recognizing the unique needs and experiences of caregivers and patients, empowering them to advocate for their rights and well-being, and gathering insights to identify opportunities for systemic improvement.

ACHIEVEMENTS

- **The Diversity, Equity, and Inclusion (DEI) Organizational Readiness Quiz** was produced and disseminated to provide additional follow up from the Patient Advocacy Organizational Capacity Survey. The readiness quiz is designed to help you and your organization to implement DEI strategies and tactics to ensure you serve all patients, caregivers, and communities.

- **RDDC hosted its 2nd Annual event, "The Eve of Rare Disease Day Reception".** This intimate event brought together nearly 70 attendees, including rare disease patients, caregivers, advocacy organizational leaders, government representatives, and industry professionals.

  - The event included an award ceremony where nine rare disease champions were presented with inaugural RISE awards. RISE stands for Resilient, Impassioned, Strong, and Empowered. The awards highlighted members and organizations of the rare disease community who encompassed the RDDC mission and led the charge for rare disease diversity advocacy:
    - **Genesis Jones**, Patient RISE Award
    - **Dr. Marshall Summar**, Provider RISE Award
    - **Sarita Edwards** - Caregiver RISE Award
    - **Patient Advocate Foundation** - Patient Advocate RISE Award
    - **Travere Therapeutics** - Industry Partner RISE Award
    - **Amgen Rare Disease**, formally Horizon Therapeutics - Honoree Industry Partner RISE Award
    - **The Office of Orphan Product Development at FDA** - Health Equity RISE Award
    - **Senator Cory Booker** - Congressional RISE Award
    - **Senator Tim Scott** - Congressional RISE Award

  - Rare Disease Day RDDC news article highlight:
    - In addition to RDDC specific media outreach during rare disease week, RDDC partners such as the Patient Advocate Foundation, helped to promote the importance of the work being conducted by RDDC, by highlighting RDDC in their media outreach.
      - Patient Advocate Foundation. "Patient Advocate Foundation Receives Patient Advocacy RISE Award." Patient Advocate Foundation, March 2024. https://www.patientadvocate.org/article/patient-advocate-foundation-receives-patient-advocacy-rise-award/

- **In February 2024, the Coalition distributed the Executive Summary of the Patient and Caregiver Gaps Survey.** This summary provides an overview of the initial findings and insights from the comprehensive data analysis conducted on the survey responses. The survey aimed to identify and understand the gaps and challenges faced by patients with rare diseases and their caregivers. It focused on identifying disparities in care and support between historically represented and underrepresented populations.

- Two key findings from this survey were:
  - Respondents encountered 14 potential factors that may have caused them to delay or forgo care. For seven barriers, over a quarter of the population reported that they led to delay or avoidance of care. These factors included issues related to emotional well-being, financial constraints, limitations stemming from their rare disease, worries about the Covid-19 pandemic, and concerns about interactions with healthcare providers.
  - When the survey focused on financial barriers to care, 21% of respondents indicated that during the past year, they could not afford their prescription medications, and 20% indicated they could not afford dental care. Among underrepresented respondents, substantial proportions were reporting an inability to afford care.
- Use this link to view the full version of the Executive Summary: Addressing Health Disparities in Rare Diseases through Comprehensive Data Collection

- **In Q1 2024, RDDC initiated Phase 2 of our Know Your Family History (KYFH) campaign.** The working group developed seven nationally accredited training modules for Community Healthcare Workers that encompass different aspects of the rare disease life cycle, emphasizing minority communities.
  - RDDC in partnership with Global Genes All in Rare Project, will launch the training into two underrepresented communities in the Fall/Winter of 2024. This effort will be one that highlights our efforts to directly impact rare disease communities.

- **This year, RDDC selected two new fellows**. Launched in 2023, this program is designed to offer a transformative experience to current and future healthcare providers and researchers to bridge the knowledge gap in rare diseases. The 2024 Fellows embarked on specialized fellowships with rare disease organizations spanning 6 to 9 months:
  - Eve Kakudji, a PhD student in Molecular and Cell Biology at the University of California, Berkeley, was hosted by RDDC. Eve researched de-identified data from Rare Diseases Clinical Research Network (RDCRN) for the development of a white paper.
  - Elijah Richardson, a Master of Public Health from Morehouse School of Medicine, and a PhD student at The University of Tennessee, was matched with Undiagnosed Disease Network and Harvard Medical School. Their collaboration centered on improving access in minoritized and underserved communities.

- **In May 2024, RDDC announced the Fall 2024 RDDC Rare Disease Fellowship program** will be hosted for six months starting in September. This fall program will include one fellow who will be hosted by advocacy organization Nephcure for Rare Kidney Disease. Applications for the fellowship closed May 28, 2024, and are under review. The candidate will be selected in August.

IN PROGRESS

- **A comprehensive report of the insights and findings from the Patient and Caregiver Gaps Survey** is being completed. This report will illuminate the rare disease community's unmet needs and challenges, as well as provide recommendations and next steps for organizations to improve support, resources, and access. Distribution of the final report will take place in Q3/Q4.
- **RDDC has partnered with the Rare Genomes Project (RGP)** at the Broad Institute of MIT and Harvard to promote a patient-driven research study led by genomics experts and clinicians who believe that the latest advances in genomic sequencing are changing medicine and should be accessible to families with rare and undiagnosed conditions. The study is currently ongoing and the genetic information generated will be shared with other scientists to ensure it has the widest possible impact.
- **RDDC is committed to educating and engaging with healthcare providers** as they are critical to bridging the gap in patient outcomes. The launch of an accredited Continuing Medical Education (CME) training program focused on rare diseases, health equity, cultural competency, and cultural humility is set to commence in August and aims to equip healthcare professionals with the knowledge and skills necessary to provide more inclusive and equitable care for patients with rare diseases. Participants will receive CME credits upon completing the program, and a certificate of completion will be provided to participants who meet all course requirements.
- **Co-sponsored the 3rd Rare Health Equity Forum** in San Diego with Global Genes. The 2023 RDDC Fellows presented on their projects and shared their experience in deepening their knowledge on rare diseases. We plan to participate in the 4th event in Kansas City in September 2024.

WORKGROUP:
Government Regulation, Legislation and Policy

The RDDC's government regulation, legislation, and policy workgroup are indispensable in confronting the many challenges underserved populations and their families face due to social determinants of health. A central objective of this workgroup is to advocate for regulatory measures within the national rare disease policy framework to meet these communities' distinctive needs. Our dedicated efforts encompass the meticulous crafting of policies and legislative initiatives to address the full spectrum of diagnosis, treatment, care, and support concerns.

ACHIEVEMENTS

- **During Rare Disease Week, RDDC was honored to present RISE awards to Congressional leaders** who have demonstrated exceptional dedication and leadership in advocating for rare diseases. This year's honorees included Senator Cory Booker of New Jersey and Senator Tim Scott of South Carolina. These awards recognize their tireless efforts in championing policies that improve access to care, research funding, and patient support services to serve as a beacon of hope for the rare disease community. By publicly acknowledging their contributions, RDDC aims to inspire other Congressional leaders to follow their example and continue pushing for legislative measures that positively impact individuals affected by rare diseases.

- **The workgroup emailed an electronic copy to all members of the Rare Disease Caucus** with an overview of the Patient Advocacy Organization's Diversity, Equity, and Inclusion Report and invited them to the Rare Disease Day Reception in Washington, DC. Staying in communication and building relationships with key members of the 118th Congress is a critical part of the workgroup's overall strategy along with advocating for policies, addressing constituent concerns, and supporting the legislative process. Being the sole coalition dedicated to historically marginalized populations impacted by rare diseases, the data we gather can equip lawmakers with the information required for informed decision-making that ultimately serves the greater public good.

- **RDDC was pivotal in advocating for the Accelerating Kids Access to Care Act**, urging Congress to pass legislation that would significantly enhance access to critical pediatric care for children with rare diseases. This legislation proposes to streamline the process for out-of-state providers to enroll as participating providers in state Medicaid programs, alleviating burdensome screening requirements that often delay essential healthcare services. This act represents a lifeline for millions of children with rare diseases who require specialized care not available locally, ensuring timely access to the expertise and treatments they urgently need. By supporting this legislation, RDDC underscored the importance of fostering a healthcare system that prioritizes accessibility and equitable treatment options for all children, regardless of their geographic location or medical condition.

- **RDDC played a key role in shaping the future of clinical trials** by actively participating in the public discourse surrounding the FDA Draft Guidance for Decentralized Clinical Trials. Our comments offered valuable insights into rare disease patients' unique challenges in decentralized trial settings. By advocating for patient-centered approaches and emphasizing the importance of diversity in trial participation, RDDC contributed to developing more inclusive and effective trial methodologies.

- **RDDC took a proactive stance in advocating for the rights of individuals with rare diseases** who have physical or mobility impairments by submitting public comments to the Department of Transportation (DOT) on their Notice of Proposed Rulemaking (NPRM) concerning accessible travel. This NPRM addressed critical issues related to the accessibility of transportation services, particularly for those using wheelchairs. By highlighting the challenges faced by millions within the rare disease community who also have physical disabilities, RDDC emphasized the urgent need for comprehensive regulations that ensure equitable access to transportation options.
- **In collaboration with the Paralyzed Veterans of America, RDDC took a significant step forward in advancing the rights of individuals** with rare diseases and building cross-sectional advocacy partnerships by signing their advocacy letter. By supporting this letter, RDDC reinforced the importance of inclusive travel policies that accommodate the unique requirements of individuals with rare diseases, thereby enhancing their ability to participate fully in society.
- **RDDC proudly joined Travere Therapeutics in signing a letter aimed at raising awareness and advocating for improved outcomes for individuals with rare kidney diseases within Asian American, Native Hawaiian, and Pacific Islander (AA/NHPI) communities.** This collaborative effort was instrumental in highlighting the unique challenges faced by these populations, who are disproportionately affected by rare kidney diseases yet often underserved in terms of healthcare access and resources. By supporting this initiative, RDDC amplified the voices of those within AA/NHPI communities, advocating for increased research, culturally competent care, and targeted interventions to address the specific needs of individuals with rare kidney diseases.
- **RDDC coalition members engaged with several Congressional staff members** from Republican and Democratic offices to introduce the Council's mission and objectives. These interactions served as a platform for RDDC to better understand the healthcare priorities of various Congressional offices. By fostering these relationships, RDDC has positioned itself as a trusted resource for lawmakers seeking insights into the challenges the rare disease community faces.

IN PROGRESS

- **RDDC has been actively mobilizing support for extending the Rare Pediatric Disease Priority Review Voucher (PRV) Program**, scheduled to expire on September 30, 2024. This vital program incentivizes the development of treatments for rare pediatric diseases by granting a voucher that expedites the review of a future drug application. By fostering these relationships, RDDC has positioned itself as a trusted resource for lawmakers seeking insights into the challenges the rare disease community faces.
- **The Workgroup's unwavering commitment** is to build and nurture relationships with key Congressional leaders from both sides of the aisles in healthcare. These connections are not just important, they are instrumental in advancing policies that specifically address the needs of rare disease patients. As RDDC gains credibility and influence, these relationships will play a crucial role in increasing awareness and support for rare disease-related initiatives on Capitol Hill.
- **Analyzing Congressional and Regulatory Policies:**
  - The Workgroup continues its meticulous analysis of congressional and regulatory policies, seeking areas of alignment with RDDC's mission. The forthcoming Patient and Caregiver Gaps Survey Report will be a powerful tool for identifying opportunities to advocate for policies that benefit rare disease patients, promote diversity in healthcare, and advocate for inclusive clinical trial practices.

WORKGROUP:
Diversity in Clinical Trials

Clinical trials of potential treatments for rare diseases are vital to identifying effective patient interventions. Given considerable health disparities in rare diseases, it is vital that including diverse populations is prioritized in the recruitment and selection of clinical trial participants[3]. This commitment stems from the understanding that clinical trials must accurately reflect the heterogeneous nature of rare disease patients, ensuring the safety and effectiveness of treatments once approved. RDDC's focus on clinical trials and the equitable inclusion of historically marginalized populations sets us apart and positions us as a beacon of change and progress.

3 Informed DNA "The importance of achieving diversity in rare disease clinical trials". April 20, 2021. Retrieved from https://informeddna.com/blog-achieving-diversity-in-rare-disease-clinical-trials/

ACHIEVEMENTS

RDDC's 2024 Rare Disease Day toolkit included the availability of GeneClips clinical trials informational videos. These videos empower patients and caregivers with the knowledge to make informed decisions. These educational resources, both in English and Spanish, comprehensively address a range of critical topics related to clinical trials, have been disseminated through the RDDC YouTube Channel, and are integrated into the RDDC website. Topics include:

- What is the difference between Clinical Genetic and Research Genetic Testing?
- Institutional Review Board and Your Protection
- Understanding Consent for Clinical Trials and General Research Studies
- Informed Consent

- **This year, RDDC has presented at various conferences**, such as the Center for Healthcare Innovation's 12th Annual Diversity, Inclusion, & Health Equity Symposium, the 28th Clinical Trial Innovation Programme, Outsourcing in Clinical Trials (article above), and the inaugural Clinical Trials in Rare Diseases Conference.

IN PROGRESS

- **A subgroup of the diversity in clinical trials and research working group is in the final stages of drafting a comprehensive review of the Rare Disease Clinical Resource Network (RDCRN) diversity demographics data** collected from clinical trial sites associated with the National Institutes of Health (NIH) over the past 20 years. This review aims to provide critical insights into the representation and participation of diverse populations in rare disease clinical trials, highlighting trends, gaps, and a baseline of information for future reference. This review is a critical step toward understanding and addressing the disparities in clinical research participation. The writing group comprises select RDDC coalition members and select members of RDCRN who will Identify appropriate journals and platforms for publication.
- **RDDC's collaboration with the Indo US Rare Disease Organization** is focused on creating a Diversity, Equity, Inclusion, and Access (DEIA) and Rare Disease white paper, which is in its final stages of development. This pivotal document aims to shed light on the intersection of DEIA principles and the challenges faced by individuals with rare diseases. This peer-reviewed paper seeks to identify gaps in the experience of diverse populations, including patients of all races and ethnicities with rare diseases. The manuscript will include facts, figures, and recommendations. The anticipated completion of this project is September 2024. Due to the complex nature of this project, RDDC was able to help our partner build their talent team by hiring additional team members to support this project and future projects.

Featured Publications:

These publications serve as a testament to the quality and relevance of the work, demonstrating thought leadership, expertise, and influence. We present a selection of our most notable publications, showcasing our commitment to excellence and innovation. They have been carefully chosen for their impact, significance, and relevance to current trends and challenges in our field. By sharing these works, we aim to demonstrate our ongoing dedication to advancing knowledge, fostering collaboration, and making a meaningful impact in our community and beyond.

- Ra, Justine. "Overcoming the barriers to diverse and inclusive patient recruitment." Clinical Trials Arena, 23 May 2024. https://www.clinicaltrialsarena.com/news/overcoming-the-barriers-to-diverse-and-inclusive-patient-recruitment/
- Vilakazi, Lindiwe. "Bringing Awareness to the Plight of Rare Disease Patients." Washington Informer, 28 February 2024, https://www.washingtoninformer.com/rare-disease-access-care/
- Black Women's Health Imperative. "Rare Disease Diversity Coalition to Host 2nd Annual Rare Disease Day Reception." PR Newswire, 14 February 2024, https://www.prnewswire.com/news-releases/rare-disease-diversity-coalition-to-host-2nd-annual-rare-disease-day-reception-302062026.html?tc=eml_cleartime
- Kahn, Hope. "Fueling Diversity in Rare Disease Research." National Press Foundation, 15 November 2023, https://nationalpress.org/topic/rare-disease-diversity-coalition-equity-debbie-drell-linda-goler-blount/
- U.S. Representative Mike Kelly. (2023, October 27). *Reps. Kelly, Matsui, Dunn, Thompson introduce legislation to support access to evidence-based care for rare disease patients* [Press release]. https://kelly.house.gov/media/press-releases/reps-kelly-matsui-dunn-thompson-introduce-legislation-support-access-evidence
- Arena International Events Group. (2023, October). *Outsourcing in Clinical Trials Handbook 2024*. Verdict. https://content.yudu.com/web/44mp6/0A44qhr/OCTHandbook2024/html/index.html?page=52&origin=reader&utm_medium=email&_hsmi=2&_hsenc=p2ANqtz-_abirZxnJFf9p99lK-P21Ww-cqOkTdFNUF3Q7B9Ru3OKhbTKLOI-KQ0-zzVyhX3APVSo2mr82jpZf-NgLu3dLlIOpjweO5ptZzTHvmlOE3WUKq4Iw&utm_content=2&utm_source=hs_email

CONCLUSION

RDDC is committed to making a lasting impact on the lives of rare disease patients and their families, particularly those from diverse backgrounds. Our progress in 2024 is a testament to the collective effort and dedication of our team, partners, and supporters.

We thank you for your partnership this year. As we look ahead to 2025 and beyond, we are excited to continue advancing rare disease awareness, education, and diagnosis with partners like you.

If you are interested in our mission and want to get involved, please email RDDC at rddc@bwhi.org.

RDDC™

RARE DISEASE DIVERSITY COALITION

www.rarediseasediversity.org