



 **RDDC**   
RARE DISEASE DIVERSITY COALITION

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# 2023 ACHIEVEMENT REPORT



**BLACK WOMEN'S**  
HEALTH IMPERATIVE



RARE DISEASE DIVERSITY COALITION

## Working Groups Leadership

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- Linda Goler Blount
- Dr. Marshall Summar
- Veronica Moore
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### **Provider Education & Engagement**

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### **Patient & Caregiver**

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### **Government Regulation, Legislation & Policy**

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## 2023 RDDDC™ Achievement Report

The Rare Disease Diversity Coalition™ (RDDDC™) actively unites influential 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 substantially impact an estimated 25-30 million individuals across the United States.

In our efforts to address the myriad challenges faced by patients afflicted with the approximately 7,000 known severely debilitating or life-threatening diseases, we must recognize and embrace the remarkable diversity within rare diseases. According to extensive research, 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.<sup>1</sup>

2023 marks the third anniversary of RDDDC and its dedicated efforts in catalyzing transformative change across policy, patient advocacy, and healthcare spheres. Our unwavering commitment remains focused on fostering equitable access to treatment for rare diseases, particularly among underrepresented populations. We firmly believe in amplifying the voices and addressing the unique needs of these individuals, ensuring that they are heard and truly understood.

RDDDC's accomplishments encompassed a wide range of meaningful actions, such as collecting data on the lived experiences, disproportionate burden, impact, and outcomes of rare diseases on and for patients from diverse racial and ethnic backgrounds and their families and communities. Additionally, we have established strong connections with influential individuals in Congress, fostering important relationships to further our cause. Furthermore, we have made great strides in expanding the RDDDC Fellowship program, allowing us to support even more aspiring leaders. Lastly, our efforts to promote the *Know Your Family History* toolkit increased engagement, empowering individuals to take control of their health.

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

## 2023 KEY ACHIEVEMENTS

In the rapidly evolving landscape of 2023, a multitude of key achievements have marked significant milestones across the Coalition:

- Created a successful pilot phase of the *Know Your Family History (KYFH)* Toolkit promotion campaign, spanning from October to December 2022. Our organization has successfully attained exceptional outcomes throughout this designated time frame, surpassing the threshold of 10,000 page views. We are pleased to announce that this exceptional performance has exceeded our initial goal by an extraordinary margin of 1000%.
- Launched the Patient and Caregivers Barriers of Access Survey, resulting in approximately 3,000 responses, majority from the historically underrepresented populations.
- Produced and disseminated a comprehensive report on the diversity, equity, and inclusion (DEI) efforts in rare diseases about the capacity of patient advocacy organizations (PAO) report. This report provides valuable insights into the efforts made by rare disease organizations in promoting diversity, equity, and inclusion. In addition, the information collected has garnered significant attention and positive feedback from stakeholders, further solidifying our commitment to fostering a more inclusive and equitable rare disease community.
- Piloted a highly successful RDDC Fellows Program with two Fellows hosted by two patient advocacy organizations. A groundbreaking initiative positioning RDDC as a leader in cultivating emerging talent and driving meaningful change within the rare disease community.
- Launched the RDDC Rare Health Equity Conference Scholarship Program for students and early career professionals. This meticulously crafted program caters to the needs of aspiring students and early-career professionals and is dedicated to cultivating inclusivity and empowering future leaders in rare health equity.
- Co-sponsor of the distinctive 3rd annual Rare Health Equity Forum scheduled for September 2023. This forum brings together experts, advocates, and stakeholders from various fields to discuss innovative strategies and collaborative efforts in addressing the unique challenges faced by individuals with rare diseases. As co-sponsors, we have helped plan the agenda, identified speakers who promoted the conference and will speak at the forum.
- Hosted an inaugural RDDC signature event in Washington, DC as an integral part of the highly regarded Rare Disease Week. The highly anticipated occasion, aptly named "The Eve of Rare Disease Day Reception," witnessed an exceptional level of attendance as over 30 distinguished rare disease PAOs, industry partners, and esteemed health equity organizations graced the event with their presence.
- Redesigned the RDDC website for easier navigation. The website has been enhanced with a visually captivating design and optimized content, ensuring seamless navigation for visitors seeking valuable insights into rare diseases and the impactful endeavors of RDDC.

## Workgroup: Delay in Diagnosis

One of the most pressing challenges of rare diseases is that their presentation can be atypical, leading to missed or delayed diagnoses<sup>2</sup> due to the complex and often atypical symptomatology associated with them. RDDC recognizes the urgency of addressing this issue, particularly among patients from diverse backgrounds disproportionately affected by misdiagnosis or late diagnosis.

Delays in accurate diagnosis can have devastating effects. RDDC believes addressing doctor-delayed diagnoses for patients with rare diseases is crucial. Addressing the issue for patients from diverse backgrounds is even more urgent: several studies indicate that patients of color, both men and women, have a significantly higher chance of misdiagnosis or late diagnosis than other patients.<sup>3</sup> In overcoming the problem of delayed diagnosis, genetic testing and access to data about the patient's family history are critical. In 2023, RDDC rolled out the next phase of the *Know Your Family History (KYFH)* toolkit to patient advocacy organizations to promote within their network. It also supported designing an educational version for community healthcare workers to incorporate into their operating structure.



## ACHIEVEMENTS

- In Q1 2023, RDDC placed significant emphasis on distributing our KYFH toolkit. The response exceeded our expectations, with over 10,000 views on the toolkit landing pages in English and Spanish.
  - The English landing page:
    - ◆ 4,957 Pageviews
    - ◆ 63% of page traffic from Facebook ads
    - ◆ 18% of page traffic from Google ads
    - ◆ The average time spent on the page was 1m 22sec
  - The Spanish Landing page:
    - ◆ 5,626 Pageviews
    - ◆ 96% of page traffic from Facebook ads
    - ◆ ~2% of page traffic from Google ads
    - ◆ The average time spent on the page was 1m 08sec
  - These engagement metrics suggest that visitors actively engage with the toolkit content rather than merely skimming through it.
- The KYFH Facebook ads campaign:
  - Thousands of clicks per ad and achieved an effective and economical cost per click that was under a dollar per click for every ad.
  - We aimed for a conversion rate (i.e., the number of people that see the ad and then click on the link) of 5% or higher, and our campaign consistently achieved a click rate ranging from 5-8% depending on the Google ad.
- Disseminating the KYFH to additional communities
  - RDDC worked closely with a member organization, Expecting Health, to distribute *KYFH* toolkit promotional materials to four grant recipients.
  - This strategic collaboration extended our reach to new communities and increased awareness about the importance of family history in rare disease diagnosis.

## IN PROGRESS

- To expand the *KYFH* toolkit reach as a resource for historically underserved populations, RDDC has partnered with Global Genes All in Rare Project to convert the *KYFH* campaign into a training module for community health care workers.
  - The working group will design a comprehensive learning and teaching module for community healthcare workers to empower them with the tools and knowledge to recognize the significance of family history in diagnosing rare diseases.
- RDDC is actively pursuing partnerships to identify free or low-cost genetic testing options for individuals at risk of rare diseases. This critical initiative will enhance early diagnosis and intervention, reducing the impact of delayed diagnoses on patients' lives.

## Workgroup: Provider Education & Engagement

Limited awareness and knowledge of rare diseases among healthcare providers contribute to doctor-delayed diagnoses and misdiagnoses. During their education, upcoming physicians receive training emphasizing prioritizing common illnesses over rare ones during patient assessments. Despite the inevitability that most physicians will encounter rare disease cases in their professional journey, many assume they will never meet these patients. This trend is especially true for primary healthcare providers such as pharmacists and pediatricians – professionals who are the initial point of contact for patients experiencing rare disease symptomatology. The tight constraints of time coupled with the sheer number of patients they manage daily further present additional obstacles.<sup>4</sup>

Furthermore, there is very low awareness of health inequities in rare diseases among health care professionals.<sup>5</sup> RDDC believes that achieving equity in the rare disease space demands expanding educational content and channels on how rare diseases impact underserved populations and inspiring the next generation of medical professionals of color to pursue careers in rare diseases.





## ACHIEVEMENT

- In Q1 2023, RDDC launched the RDDC Fellows Program, a transformative initiative designed to bridge the knowledge gap among healthcare providers regarding rare diseases. Two dedicated RDDC Fellows embarked on specialized fellowships spanning 6 to 9 months:
  - Dr. Aditi Kantipuly, an early-career physician with a unique blend of government, academic medicine, and journalism experience, collaborated with Dr. Connie Lee and the Alliance to Cure Cavernous Malformations. This partnership facilitated Dr. Kantipuly's engagement in genetics, health equity, and sustainable community-based care.
  - Rewaa Elgazar, a pre-medical Molecular & Cellular Biology student with personal experience with Multiple Sclerosis, joined forces with Dr. Rhonda Cady at Gillette Children's Specialty Healthcare. Their collaboration centered on stem cell-based therapies, health advocacy, and addressing disparities.
- RDDC Fellow curriculum is a self-paced rare disease learning module series featuring essential topics such as: rare disease fundamentals, policy and regulation, cultural competence, cultural humility, and disparities in clinical trial diversity. Subject matter experts and healthcare providers contributed their expertise to shape these comprehensive modules.

## IN PROGRESS

- In May 2023, RDDC announced a new application cycle for the RDDC Rare Disease Fellowship program. Hosted for six months starting in September 2023, the program aims to nurture emerging talents and drive rare disease knowledge across diverse disciplines. Our two esteemed host sites, the Global Foundation for Peroxisomal Disorders and the Undiagnosed Diseases Network are ready to welcome the next cohort of RDDC Fellows. Applications closed on June 30, 2023, and are currently under review by the working group.
- In May 2023, RDDC announced a new Rare Disease Health Equity Conference Scholarship Program. This initiative empowers medical and allied health professionals and students committed to equity, diversity, and inclusion. Ten scholarships, each valued at \$2,000, will be awarded to individuals contributing to the field through insightful essays or videos summarizing conference learning. By providing these scholarships, we aim to introduce early-career providers and professionals of color to rare disease and health equity learning opportunities in the hopes that they will pursue or further their careers in providing healthcare to patients of the Rare community. The Call for Applications closed on August 31, 2023; the chosen recipients will have the opportunity to enhance their knowledge and contribute to the rare disease community.
- RDDC is making strides in fostering cultural competence and humility among care providers. A current partnership is creating a Continuing Medical Education (CME) module to highlight the importance of providing equitable health treatment for rare disease patients. The project is on track for launch in Q1 2024.
- Building on our commitment to diversity in healthcare, RDDC allocated funds to engage student medical organizations in initiatives that bolster diversity in medical schools and promote learning opportunities related to rare diseases and health equity. RDDC has initiated discussions with potential partner organizations to drive this essential initiative forward.

## Workgroup: Patient and Caregiver

Rare disease patients and their caregivers are at the heart of RDDC's mission. To serve these populations better, 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 underappreciated. 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 Patient Advocacy Organizational Capacity Survey, in collaboration with UpEquity, was analyzed and produced into a report for distribution at the beginning of Q1 2023. Key highlights from the report include:
  - Organizations with 2-9 employees or volunteers demonstrated increased efforts in DEI initiatives.
  - Respondents expressed the need to collect demographic information strategically to inform more substantial DEI efforts.
  - Organizations are actively initiating changes but expressed dissatisfaction with their current progress.
  - Higher-level employees displayed less favorable perceptions of their organizations' commitment to DEI.
  - Educational materials and website updates were identified as effective means to promote DEI efforts.
- RDDC hosted its inaugural signature event, *The Eve of Rare Disease Day Reception*, bringing together 50 attendees, including rare disease patients, caregivers, advocacy organizations, government representatives, and industry professionals. The event provided a platform to share insights from the Organizational Capacity Survey and foster collaboration.
- The Patient and Caregiver Gaps Survey collected almost 3,000 responses, predominantly from historically underrepresented populations. The survey aims to highlight the challenges patients and caregivers face, giving voice to their experiences.

## IN PROGRESS

- For the Patient and Caregiver Gaps Survey work is ongoing to analyze the data collected. We will compile the insights and findings into a comprehensive report, which we plan to distribute in Q3-Q4. This report will illuminate this essential community's unmet needs and challenges.
- During the National Organization for Rare Disorders (NORD®) Rare Diseases and Orphan Products Breakthrough Summit in Oct 2023, we will release the preliminary results of the Gap Survey, ensuring broader dissemination of valuable insights.
- To support PAOs in their DEI initiatives, RDDC is developing a toolkit. This resource will provide actionable recommendations from the PAO survey findings, offering practical steps to drive meaningful and systemic organizational change.

## Workgroup: Government Regulation, Legislation and Policy

The RDDC's government regulation, legislation, and policy workgroup are indispensable in confronting the myriad 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 tailored 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

- **Informing and educating the Rare Disease Caucus.**

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. The workgroup sent this report as a crucial means of communicating information to key members of the 118th Congress, advocating for policies, addressing constituent concerns, and supporting the legislative process. Being the sole coalition dedicated to rare diseases and historically marginalized populations, the data we gather can equip lawmakers with the information required for informed decision-making that ultimately serves the greater public good.
- **FDA Draft Guidance on Decentralized Clinical Trials.**

RDDC played a pivotal 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.
- **Relationship development with Congressional staff members.**

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 specific challenges faced by the rare disease community.

## IN PROGRESS

- **Cultivating Relationships with Congressional Health Care Leaders.**

The Workgroup is committed to building and nurturing relationships with key Congressional leaders from both sides of the aisles in healthcare. These connections are instrumental in advancing policies that address the specific needs of rare disease patients. As RDDC gains credibility and influence, these relationships will increase 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 results 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 population is prioritized in the recruitment and selection of clinical trial participants.<sup>6</sup> 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. RDCC'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.

In 2023, RDCC engaged in several initiatives designed to address these obstacles.



## ACHIEVEMENTS

- To further advance education on clinical trials in English and Spanish. A collaborative effort with RDDC has yielded the availability of GeneClips clinical trials informational videos. These videos empower patients and caregivers with the knowledge to make informed decisions. These educational resources comprehensively address a range of critical topics related to clinical trials, have been strategically disseminated through the RDDC YouTube Channel, and are seamlessly integrated into the newly launched 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
- As of December 2022, RDDC is a proud partner with Bio New Jersey. As a Supporting Partner, RDDC supports the Health Equity in Clinical Trials Business Case Competition, part of the BioNJ Health Equity Initiative, and will be featured as an In-Kind Contributor on promotional materials, participate as a judge at the competition, and serve as a steering committee member. As a result of this partnership, RDDC participated in BioNJ's Inaugural *Health Equity in Clinical Trials MBA Business Plan Case Competition* on December 5, 2022, at Rutgers University, where eight teams from MBA programs developed strategies to increase diversity in clinical trial participation. The competition highlighted the challenges faced by people of color, comprising only 16% of research participants. The winning teams focused on increasing diversity in Alzheimer's trials, harnessing data for health equity targets, and boosting Hispanic/Latinx participation by providing training to community healthcare workers.
- RDDC's 2023 Rare Disease Day toolkit included two podcasts: *Workforce Diversity in Clinical Trial Research* and *Cultural Competencies and Health Literacy within the Healthcare System*. These podcasts delve into critical aspects of clinical trials, from workforce diversity's influence on trial participation to the impact of cultural competencies on patient care. These topics address the unique challenges of rare diseases and their intersection with historically marginalized populations.
- On March 21, 2023, at the Rare Disease Innovation & Partnership Summit, RDDC moderated an expert panel discussing integrating racial disparity into the conversation about rare diseases, highlighting the impact of lack of representation in clinical trials on health outcomes, and exploring solutions to raise awareness and progress towards an equally accessible system.

## IN PROGRESS

- Beginning Q4 2023, NIH's Rare Disease Clinical Research Network (RDCRN) will provide data on the participation of people of color in rare disease clinical studies. This information will contribute to research, evaluation, and confirmation of the composition of the patient base in rare disease clinical trials. RDDC will analyze the information and document the findings for distribution in 2024.
- RDDC's commitment to increasing clinical trial awareness and participation continues through patient-focused conference talks:
  - Beginning in September 2023, RDDC will moderate the RARE Health Equity Forum panel. The panel will focus on improving patient participation in clinical trials and research by addressing barriers at multiple levels and identifying strategies focused on engaging participants from low-income and communities of color.
  - The working group continues to explore partnerships with Rare Disease Advocacy Groups to design strategies that educate, recruit, and encourage participation from high unmet need populations.
- As of July 2023, RDDC's collaboration with Indo US Rare Disease Organization is yielding a whitepaper, peer-reviewed article, and reporting template to define Diversity in Rare Disease Research and Clinical Trials baselines. The initiative aims 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 based on the anticipated completion of this research portion in May 2024.
- In August 2023, to increase clinical trial (CT) awareness among low-income and communities of color, the working group will review opportunities to partner with Rare Disease Advocacy Groups, such as Nephcure, The Alliance to Cure Cavernous Malformations, National PKU Alliance, Undiagnosed Diseases Network, Guthy-Jackson, Couch Pennies, and Accelerated Cure Project, to design and implement a strategy to educate, recruit and encourage participation in CTs from high unmet need populations. This initiative will extend into 2024 to continue enhancing our impact on underserved populations.



## 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 2023 is a testament to the collective effort and dedication of our team, partners, and supporters.

As we look ahead, we are excited to continue advancing rare disease awareness, education, and diagnosis with partners like you.



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