BLACK WOMEN'S
HEALTH IMPERATIVE

RDDC
RARE DISEASE DIVERSITY COALITION

2022 RDDC Achievement Report

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The Rare Disease Diversity Coalition (RDDC) was convened to bring together key stakeholders to tackle the extraordinary obstacles and challenges faced by rare disease patients of color, their families and communities.

Although individual rare diseaases are by their very definition rare, considered cumulatively, having a rare disease is common, affecting an estimated 25-30 million people across the US. At the same time, there is considerable diversity in the population of patients affected by the 7,000 or so known severely debilitating or life-threatening diseases, making it crucial to address diversity in rare diseases.[1] Research shows, however, that minority groups and communities of color experience considerable health disparities when they are affected by a rare diseases. Minority patients 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 that disproportionately affect communities of color are less likely to attract funding for research.

Many other barriers to equity and inclusion in rare diseases exist. For instance, there continues to be underrepresentation of minority groups among clinical trial participants and in the data stored in rare disease databases and registries, which undermines advancement in knowledge of how rare diseases develop, progress and should be treated in diverse populations. Overall, more tangible work is needed to recognize 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.

Two years into the Covid-19 pandemic, 2022 saw RDDC continue to drive change at the policy, patient advocacy and healthcare levels to ensure that patients from all backgrounds, races and walks of life have equitable access to treatment for rare diseases, and that their needs are heard and understood. Our accomplishments were many, including contributions to new and upcoming legislation, raising awareness of disparities in access to treatment and insurance coverage for rare diseases, providing education on health equity to medical health professionals and trainees and building knowledge of the need for diversity in clinical trials.

Moving forward, the RDDC continues to fight to eliminate the obstacles that communities of color face daily in obtaining timely diagnoses, information and access to effective and quality treatment for rare diseases.

2022 KEY ACHIEVEMENTS

- **1 major marketing campaign in development**
- **2 significant pieces of legislation in development**
- **5 Webinars held**
- **1,179 Webinar attendees**

Workgroup: Delay in Diagnosis

One of the defining characteristics of rare diseases is that their presentation can be atypical, leading to missed or delayed diagnoses[2]. Symptomology can be complex, obscure, multisystem and unusual, and since they typically do not correspond with known medical conditions, can lead to inaccurate, contradictory or delayed diagnoses, or even accusations of malingering.

Delay in accurate diagnosis can have devastating effects. In addition to delaying access to the critical treatment that patients need, while rare disease patients are awaiting accurate diagnosis, they may experience worsening of their symptoms, and some even die. Diagnostic delay is also known to have a significant effect on patients' psychological health and wellbeing and their quality of life, leading to clinically significant levels of mental anguish, depression, sense of isolation, feelings of doom and anxiety. Their unmet informational needs can reduce their level of trust in the medical establishment and decrease their satisfaction with care.

For these reasons, RDDC believes it is crucial to address doctor-delayed diagnoses for patients with rare diseases. For patients from diverse backgrounds, addressing the issue is even more urgent: several studies indicate that patients of color, including women, have a significantly higher chance of misdiagnosis or late diagnosis than other patients[3]. In overcoming the problem of delayed diagnosis, genetic testing and access to data about the patients' family history are key. That's why, in 2022, RDDC has focused on rolling out the *Know Your Family History* campaign and begun work on identifying free or low-cost genetic testing options for patients.

ACHIEVEMENTS

- Campaign design, product deliverables and communications/partnership strategy for the *Know Your Family History* campaign
- Developed a data analysis strategy for the campaign
- Begun to outline a strategy for the partnership development phase of the campaign
- Identified potential funders/partners for identifying free/low-cost genetic testing options

IN PROGRESS

- Securing strategic partnerships for the next phase of the *Know Your Family History* campaign through marketing
- Continue to pursue partnerships for identifying free/low-cost genetic testing options

Workgroup: Provider Education

One of the reasons for doctor-delayed diagnoses and misdiagnoses is the low awareness and knowledge of rare diseases among healthcare providers[4]. During their studies, future physicians are trained to first consider common and not rare diseases when evaluating patients, and although most physicians will actually encounter (through diagnosis or treatment) a rare disease patient in their career, most assume that they will never meet these patients. This is especially the case for primary healthcare providers such as pharmacists and pediatricians – professionals that are the first port of call for patients experiencing rare disease symptomology. Time constraints and the sheer number of patients that primary care providers encounter on a daily basis act as additional obstacles.

Furthermore, there is very low awareness of health inequities in rare diseases among health care professionals. RDDC believes that achieving equity in the rare disease space demands that education of medical professionals includes recognizing rare disease symptoms and that treating patients with rare diseases must be accompanied by education on how rare diseases impact communities of color.

ACHIEVEMENTS

- In the fall of 2021, RDDC partnered with the National Organization for Rare Disorders, Inc. (NORD) to develop a series of three webinars focused on identifying health inequities in the rare disease space and exploring the role that healthcare professionals (HCPs) can play in effectively addressing these issues. The webinars - *Health Equity and Rare Disorders, Advancing Equity in Rare Healthcare* and *Walk in our Shoes: The Experience of Rare Disease Patients* aired in December 2021, February 2022, and April 2022, attracted a total of 1,179 participants, including HCPs, medical students, patient advocates, industry, and academics and received positive feedback from attendees.
- In April 2022, the inaugural Rare Disease Fellowship program was announced, aimed at inspiring the next generation of the medical workforce to work in the rare disease space and achieve greater health equity for people of color living with rare diseases. The Fellowship is open to undergraduates, medical students, early-career physicians, nurses, medical researchers who have been practicing less than five years, and medical or nursing residents who have a demonstrated interest in addressing rare disease disparities among communities of color. Each Fellow is hosted for a period of 6-9 months at an organization specializing in rare disease. 16 interested participating organizations were narrowed to 3 finalists, each of is being matched to a selected Fellow in August-September 2022.

IN PROGRESS

- Applications for potential fellows are being received and will be reviewed after the deadline ends. The curriculum for the fellows is currently being developed.

Development of a CME module by NMA and NHMA for health care providers to learn about rare diseases and how to rare disease patients with a lens on health equity.

Workgroup: Patient and Caregiver

Rare diseases impact diverse populations, and in order to serve those populations better, it is vital that we gather data on the experiences of diverse patients with rare diseases, and their caregivers. For instance, research suggests that the vast majority of patients with rare disease take responsibility for coordinating their own care, or do not have comprehensive care plan in place[5]. Accordingly, their social lives and working schedules are likely to be significantly impacted. Gathering insight into how diverse patients experience rare diseases and their treatments is a crucial input for medical professional education, and to raise awareness of the specific challenges associated with diversity in rare diseases.

Caregivers are also affected. It is estimated that 1 in 9 workers or 1 in 5 adults is a caregiver for another person[6]. Yet, caregivers are rarely recognized as a segment of diversity. Caregivers should be understood as having specific needs and experiences, and organizations, such as employers, increasingly need to recognize those needs. That is the focus of RDDC's work in this area.

ACHIEVEMENTS

- The Organizational Capacity Survey was launched in June 2022 in collaboration with UpEquity and is designed to gather information on current efforts among rare disease patient organizations to reach diverse populations. Work to support the survey launch included survey development and development of an implementation timeframe, pilot testing, and development of a PR/communications strategy (including flyers, social media language and a press release).

IN PROGRESS

- Work is continuing on the Patients and Caregiver Gaps Survey initiative, with contractor selection now complete. The next steps will involve creating an implementation strategy for the project complete with interim deliverables, deadlines and evaluation metrics.

Workgroup: Government Regulation, Legislation and Policy

While by definition, individual rare diseases impact few people, they have a cumulative impact on public health in terms of the severity of their symptoms and the total number of people they impact. Many rare diseases have poor prognoses and few effective treatments, making nationwide research into their pathophysiological mechanisms crucial. Furthermore, the issues impacting communities of color experiencing rare diseases are so complex and multifarious (barriers in access to care, delayed diagnoses, limited knowledge among healthcare professionals of the specific issues impacting patients from diverse backgrounds), that they can only be adequately addressed through government regulatory action.

RDDC's workgroup on government regulation, legislation and policy is playing a critical role in addressing these challenges by amplifying the voices of rare disease patients of color and their families and lobbying for regulatory action on national rare disease policy to address their distinct needs. Our work has focused on the development of policy and legislation to address the diagnosis, treatment, care, and support of patients of color with rare diseases, with priority given to action that promotes evidence-based solutions to alleviate the disproportionate burden of rare diseases on communities of color, to address disparities in rare disease research, access, and care, and to advance innovation to meet unmet medical needs in rare diseases.

Through its advocacy, the Government Regulation, Legislation, and Policy working group has made significant progress and achievements on the Hill to address and find solutions for the extraordinary challenges faced by rare disease patients of color nationwide.

ACHIEVEMENTS

- The RDDC worked with policymakers to reshape the FDA User Fee legislation. In a letter to Chair Murray and Ranking Member Burr, of the Senate Health, Education, Labor, and Pensions Committee (HELP), the RDDC formally commented on the Senate FDA Safety and Landmark Advancements Act (FDASLA) discussion draft. The RDDC believes that the FDA user fee reauthorization process presents a wonderful opportunity to enact meaningful healthcare policies to accompany the direct user fee reauthorization. Also, the RDDC strongly encouraged the HELP Committee to seize this opportunity by, among other things, addressing the important need to enhance diversity in clinical trials, facilitate prompt access to treatments, and to further advance innovation to meet unmet medical needs in rare diseases. The RDDC submitted proposals regarding opportunities to make a difference on issues related to the two pillars of RDDC's mission: rare diseases and diversity/equity.

- In February 2022, Congressman G.K. Butterfield (NC-1), a Co-Chair of the Rare Disease Congressional Caucus and 24th Chair of the Congressional Black Caucus, introduced a Resolution (H. Res. 948) recognizing the extraordinary challenges faced by patients of color with rare diseases and the need to identify and promote evidence-based solutions to alleviate the disproportionate burden of rare diseases on these communities, as well as supporting Rare Disease Day. Notably, the Resolution explicitly honors the work and overall mission of the RDDC.
- Also in February 2022, Congressmen G.K. Butterfield (NC-1) and Bobby Rush (IL-1) submitted statements to the congressional record expressing support for the advocates, medical professionals, and caregivers who dedicate their lives to providing resources to those diagnosed with rare diseases. The statements recognized the RDDC's membership and initiatives.
- In March 2022, U.S. Representative Charlie Crist (D-St. Petersburg) and U.S. Representative Barbara Lee (D-California) introduced H.R. 7177, the Sickle Cell Care Expansion Act. This legislation will improve access to treatment, medical care, and quality of life for people suffering from Sickle Cell Disease (SCD). Specifically, the legislation expands the National Health Services Corps (NHSC) to include the study of benign hematology, the specialty under which SCD falls. NHSC provides scholarships and loan repayment assistance as an incentive to attract healthcare providers to specialties where shortages exist.

IN PROGRESS

- Work is ongoing with the lead sponsor and House legislative counsel to finalize language for RDDC's signature legislation, tentatively named the Elimination of Disparities in Rare Disease Research, Access, and Care Act of 2022. This signature legislation for RDDC would address disparities in rare disease research, access, and care. It also would institute programs to increase diversity in clinical trials. The provisions include 1) developing a comprehensive federal plan to address rare diseases in minority populations, 2) conduct or support funding opportunity announcements, grants, or cooperative agreements for rare diseases in minority populations, 3) institute research regarding early detection, diagnosis and treatment of rare diseases, and 4) provide clinical training programs.

Workgroup: Diversity in Clinical Trials

Clinical trials of potential treatments for rare diseases are a vital step on the journey to identifying effective intervention for patients. Given considerable health disparities in rare diseases, it is vital that diversity is prioritized in the recruitment and selection of clinical trial participants[7].

Diversity is crucial in clinical trials for several reasons. First, the safety and effectiveness of new treatments can only be discerned if they are evaluated among participants who are representative of the patient population who are intended to use the treatment, once it is approved. Rare diseases affect heterogeneous populations, and thus it is vital that clinical trials generate findings generalizable to diverse populations and identify groups where treatments may be less effective or may produce differential responses and side effects. Given mistrust in the medical establishment by some minority groups, participation of minorities in clinical trials can build confidence among communities of color in new treatments. Finally, note that clinical trials are the primary route by which rare disease patients can access unapproved investigational treatments. Thus, enhancing diversity can go some way towards addressing health inequities by providing patients of color access to potentially effective and lifesaving healthcare that might otherwise be unavailable.

For these reasons it is crucial to engage clinical trial participants who are diverse in gender, race, ethnicity and age in clinical trials.

According to TrialFacts, there are a number of barriers to enhancing diversity in clinical trials including patient mistrust in the process, lack of information about, and comfort with the process, lack of cultural competencies among study staff and logistical barriers[8]. In 2022, RDCC engaged in a number of initiatives designed to address these obstacles.

ACHIEVEMENTS

- In June 2022, the *Know Your Options: Careers in Clinical Trial Research* webinar was held, featuring a panel discussion of minority clinical trial researchers and discuss their career development and serve as a resource to provide career options to medical students of color as they consider professional alternatives. The webinar was amplified through BWHI and RDDC's existing newsletters and cross-promoted by relevant RDDC member organizations and stakeholder affiliates (National Hispanic Medical Association, National Medical Association, National Organization

of Rare Disorders, MANA National, National Coalition of 100 Black Women, National Black Nurses Association etc.) to target medical association student chapters (Student National Medical Association, NORD Students for Rare Program etc.) and clinical trial workforce networks and consortiums (Black Women in Clinical Research, Women of Color in Pharma etc., NIH) in order to maximize awareness and participation among medical students of color and junior trialists.

- In July 2022, the Rare Disease Clinical Trials Ecosystem for Patients of Color Webinar was held, featuring a panel discussion of the various professionals involved in clinical trial research to increase awareness, education, and participation of communities of color in research and clinical trials.
- Confirmed the participation of Meharry Medical College and Morgan State University at the September 2022 HBCU-HSU President Roundtable on Workforce Diversity in Clinical Trial Research.

IN PROGRESS

- Beginning Q3 of 2022, the Rare Disease Clinical Research Network (RDCRN) will begin providing data around the participation of people of color in rare disease clinical studies in order to research, evaluate, and confirm the composition of the patient base in rare disease clinical trials, and then publish the findings.
- In September 2022, the HBCU-HSU President Roundtable will be focused on Workforce Diversity in Clinical Trial Research. In this leadership roundtable, Presidents and Deans of HBCUs and HSIs will discuss the critical policies, programs, and interventions needed to inform medical students of color about clinical research opportunities. Segments from the roundtable will be developed to use as an educational tool for clinical trial researchers and medical professionals.
- Beginning Q4, 2022, the GeneClips patient app for rare diseases will offer quick and easy-to-understand educational videos that patients of color and their families can use to understand how research and clinical trials work in order to encourage greater diversity in participation.
- In October 2022, the development of a Rare Disease Patient Interviews webinar which will feature testimony and stories from diverse rare disease patients who have participated in clinical trials as well as an extensive Q&A to help attendees broaden their understanding of health equity and the rare disease community.
- In November 2022, a webinar on Diversity and Cultural Competence in Research will be offered for current and future medical professionals that are involved in all aspects of clinical trial research. The webinar will feature DEI experts and leading health equity voices on the importance of embedding cultural competency in clinical trial design, incentivizing medical students of color to pursue a career in clinical trial research and educating junior coordinators and volunteers new to the field about the importance of cultural competence and linguistic competence contextualized in a trial setting.

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