



THE RARE DISEASE DIVERSITY COALITION

Charting the Path Forward for Equity in Rare Diseases

Action Plan and Coalition Overview

Contents

Foreword	3
Executive Summary	4
Charting the Path Forward for Equity in Rare Diseases	5
Rare Diseases	5
Orphan Drugs: Not Enough Rare Disease Therapies.	5
The Orphan Drug Act and Related Policies—A History of Partial Solutions	5
Racial Health Disparities in Rare Diseases.	6
Health Disparities in America	7
A Nation Ready for Change	8
Leading the Way: The Rare Disease Diversity Coalition.	8
How Recommendations Formed	9
Top-Level Recommendations	9
A Path Forward—Change Is Possible	10
Reducing Disparities Through Administrative Action	11
Reducing Disparities Through Legislation	11
Conclusion	11
Acknowledgments	12
About BWHI	12
Steering Committee	12
Industry Sponsors	13
How to Get Involved	13
Coalition Membership.	13
Sponsorship and Industry Support	13
2021 Schedule of Meetings	13
Contact RDDC	13
Appendix 1: 2021 Actionable Roadmap	14
References	18

Foreword

It is my pleasure to present the Rare Disease Diversity Coalition (RDDC). RDDC is born out of the collective commitment of its founders to be a catalyst for progress for people of color with rare diseases. People of color with rare diseases strive to avoid being left behind in health care while feeling the effects of historic, systemic racism and its lingering disparate social, economic, and health effects. The purpose of this document is to serve as an introduction to RDDC and to describe the journey that led Black Women's Health Imperative (BWHI) and other founders to form RDDC. It is also meant to elucidate our connection to a historic coalition that stood united to demand changes in a health care system that had left rare disease patients behind in a rapidly advancing scientific revolution and our strong sense of belief—and hope—that, similarly, this coalition can help society seize the momentum for change on racial inequities and drive progress on the continuing struggles regarding health equity and rare diseases.

BWHI has been working toward progress at the crossroads of the struggles of patients of color—due to both rare diseases and health disparities—for decades. In 2020, as the devastating COVID-19 pandemic highlighted the additional struggles of patients with rare diseases and people of color, BWHI, rare disease experts, patient organizations, health and diversity advocates, and industry leaders with a deep knowledge of the medical, industry, regulatory, and cultural challenges facing minorities with rare diseases came together and established an expert steering committee, formed working groups and developed initial recommendations. As we developed the ideas underlying RDDC's mission, we have been encouraged by the momentum for change. Over the past few months, key policymakers—from the Biden Administration to Congress to state governments—have expressed the will to develop policy to address racial inequities. BWHI has been further encouraged after engaging some of these policymakers about the prospect of working cooperatively with RDDC on these issues.

We are humbled to note the similarities with the historic groups that advocated successfully for policies to change the system to incentivize inclusion of rare diseases in research and development of scientific advancements. That coalition, which resulted in the development of NORD and enactment of the Orphan Drug Act (ODA), continues to advocate successfully for patients with rare diseases. We are proud to have some of the groups involved in the successful ODA advocacy at the table in the RDDC. Beyond its inspiration and success, that earlier coalition's approach to policy solutions led to success—and is instructive on how to incentivize changes that take into account input from patients, policymakers, academic researchers, health care providers, and commercial innovators.

We hope the ideas discussed here are just the beginning. Our goal is to hone these recommendations and ideas—and to leverage the collective experience, expertise and ideas within RDDC to drive change for people of color with rare diseases. The time is now! Please join us in this journey.

Sincerely,



Linda Goler Blount, MPH

President & CEO

Black Women's Health Imperative



Executive Summary

The Rare Disease Diversity Coalition (RDDDC) formed under the leadership of the Black Women’s Health Imperative (BWHI) to help address the extraordinary challenges faced by rare disease patients of color. As advocates for patients who deal with dual struggles—to be included in the promise of scientific advancements that improve health outcomes and not be left behind because of their rare condition or their race—the leaders of RDDDC envision a unique opportunity to contribute to progress for both the rare disease and health disparities movements. This paper will describe the journey that RDDDC is taking to contribute to real and significant progress.

First is a discussion of the historical struggle of people with rare diseases, initially left behind (or “orphaned”) as scientific advances were delivering hope to people with mainstream conditions that were deemed as “commercially viable.” It then discusses how a coalition of patient advocates, researchers, health experts, and policymakers came together to advocate for changes that would ultimately lead to the Orphan Drug Act (ODA)—policy change to incentivize development of therapies for rare diseases and to overcome profitability concerns. The paper then examines the success of the ODA, the improvements that remain necessary, and one group within the rare disease community—people of color (POC)—who experience an additional struggle with health disparities that compound the barriers to health, inclusivity, and equity. For people of color who have rare diseases, the ODA’s intended course corrections have rarely found their way to deliver on the promise of scientific advancements to them.

Next, we review the shameful history of racial discrimination and its impact on people of color in every facet of society, including the social determinants of health and health disparities. While the road to equity is tough, shocking exposure of systemic racism and inequities in 2020—from policing to the disparate impact of the COVID pandemic on people of color—has contributed to the tone and momentum of a new day. Society is demanding real change to address racial inequities. RDDDC is encouraged by the discussions of individuals and institutions, including the Biden Administration and Congress, on these issues.

RDDDC is ready to lead the way on health disparities in rare disease—inspired by the advocates who served as catalysts in 1983 with ODA and continuing through 21st Century Cures. Understanding that hard work remains on rare diseases, and especially on health disparities, RDDDC convened rare disease experts, patient organizations, health and diversity advocates, and industry leaders with a deep knowledge of the medical, industry, regulatory, and cultural challenges facing people of color with rare disease. RDDDC established an expert steering committee, formed working groups, and developed initial recommendations in four areas: (1) research and clinical trials; (2) delays in diagnosis and treatment; (3) patient and provider education and engagement; and (4) policy development. The preliminary work and recommendations of these work groups is outlined below and detailed in the appendix. But this is only the beginning. Building on these plans, we will continue to develop RDDDC’s priorities and turn them into action. We are issuing an ongoing invitation to join the Coalition and contribute to its progress.

Charting the Path Forward for Equity in Rare Diseases

Rare Diseases

Today, over 30 million Americans live with a rare disease.¹ Half of those Americans are children. While the causes of the roughly 7,000 rare diseases vary, they all profoundly impact patients, their families, and our communities.² Over two-thirds of rare diseases are associated with a reduced lifespan.³ Roughly one-third of children born with a rare disease will not live to the age of five years. While some rare diseases affect only a handful of patients, the collective impact of rare diseases is a strain on families.

Orphan Drugs: Not Enough Rare Disease Therapies

“Millions of Americans who suffer from rare diseases live without hope!”⁴ That was the testimony of a patient advocacy group’s vice president to Congress in 1980. The testimonial went on to say that there was not enough spending power among patients with rare diseases to make therapeutic drug manufacture profitable. For nearly a century, our society has benefited from scientific developments that have saved and advanced the lives of many patients. During the 1970s, it became apparent that these advances were not reaching everyone. As more scientists, patient advocates, and policymakers took note and studied this issue, they realized that very few therapies for rare diseases, which became known as “orphan drugs”, had been developed, because the limited prevalence of populations with rare disease was viewed as a barrier to the commercial investment necessary to earn FDA approval.⁵ The table below reflects some of the uncertainties and costs associated with developing orphan drugs. Combined with limited post-approval profit potential, high research and development costs led to the view that the cost-benefit analysis worked against developing these therapies.⁶

	Mass-Market	Rare Disease
Large pool of patients for studies	✓	✗
Economies of scale to produce drugs	✓	✗
Large number of patients to sell to	✓	✗
Increased product liability risks ⁷	✗	✓

By the late 1970s, the issue of orphan drugs was on policymakers’ radars due to the coordinated efforts of patients, advocacy groups, scientists, and policy experts.⁸ These efforts led to a coalition that ultimately resulted in formation of the National Organization for Rare Disorders (NORD). During this time, numerous hearings were held, studies were conducted, and working groups were formed, ultimately building momentum for legislative action. These efforts led to a coalition that ultimately resulted in formation of the National Organization for Rare Disorders (NORD).

The Orphan Drug Act and Related Policies—A History of Partial Solutions

After years of pressure, on January 4, 1983, the Orphan Drug Act (ODA) was signed into law.⁹ The ODA and related policies incentivized the development of orphan drugs through tax credits, market exclusivity, grants, and waiving of Food and Drug Administration (FDA) user fees.¹⁰ These incentives offset many of the costs and risks associated with developing orphan drugs.

The ODA has made progress in its mission to incentivize the development of therapies for rare diseases.¹¹ Prior to the ODA, only 38 orphan drugs had received FDA approval; since the ODA's passage, over 900 orphan designations have been approved.¹² Further, a growing number of therapies to treat pediatric rare diseases have been approved.¹³

Since the ODA was enacted, other policies have been developed to support patients with rare diseases. The Department of Health and Human Services (HHS) has a number of administrative policies and programs designed to implement the policy of the ODA. For example, FDA's Office of Orphan Products Development administers the Rare Pediatric Disease Priority Review Voucher Program and allocates grants for rare disease-related research, clinical trials, and natural history studies. More recently, the 21st Century Cures Act has facilitated the development of rare disease drugs by modernizing the FDA approval process and by encouraging development of "targeted drugs for rare diseases."¹⁴ The expansion of newborn screening also helps patients with rare diseases. All states now universally screen newborns for at least 30 rare diseases.¹⁵ Patients diagnosed with rare diseases through newborn screening tend to live longer, manage their symptoms better, and have fewer emergent medical crises.

FDA's accelerated approval policies, which allow drugs for serious conditions that fill unmet medical needs to be approved based on a surrogate endpoint, have increased quick access to cutting-edge treatments for many rare disease patients.¹⁶ To support orphan drug development, FDA recently published guidance on "Natural History Studies for Drug Development."¹⁷ In November 2020, FDA published important guidance on "Enhancing the Diversity of Clinical Trial Populations." These policies have collectively helped the rare disease community.

The steps taken have incentivized development of therapies for rare diseases, but more work needs to be done. Over 90 percent of rare diseases still lack FDA-approved treatments.¹⁸ On average, it takes five years and seven physician consultations to accurately diagnose a patient with a rare disease.¹⁹ Once a diagnosis is made, if a treatment is approved, patients still face many hurdles to access, including cost, prior authorization requirements, and step therapy rules. More needs to be done to help the entire rare disease community.

Racial Health Disparities in Rare Diseases

While the rare disease community continues to face hurdles generally, people of color face additional hurdles in their quest for care. Barriers to diagnosis and treatment for people of color often have deadly consequences. Flaws across the entire system have a compounding effect on the care that Black, Native American, Hispanic, Asian, and Pacific Islander Americans with rare diseases receive.

Americans of color continue to be underrepresented in genome-wide association studies and clinical research trials, leading to a lack of understanding about effective treatments, particularly in diverse populations. Despite making up more than 38 percent of the U.S. population, people of color comprise only 16 percent of research study participants.²⁰ On the patient side, people of color are less likely to have affordable access to health care and rare disease experts.²¹ To make matters worse, some rare diseases disproportionately impact people of color. For instance, sarcoidosis, sickle cell anemia, thalassemia, and some forms of lupus are known to affect minority populations at higher rates than the general population.²² And implicit bias particularly harms people of color with rare diseases.²³

Health Disparities in America

A long history of discrimination and socioeconomic inequality has led to a health crisis for people of color in the United States. Both social and physical determinants of health, which disproportionately negatively affect people of color, contribute to this crisis.²⁴

Health is correlated with socioeconomic status (SES);²⁵ in general, the higher the SES, the better health a family has. In contrast, people living with lower SES generally have poorer health. Racial disparities in socioeconomic status persist.²⁶ Communities of color generally have lower SES, since the historical roots of inequality have often gone unaddressed. Environmental causes of health issues, such as lead paint poisoning, disproportionately impact poorer Americans and people of color.²⁷ Lack of adequate insurance is a major factor in hindering access to medical care for people of color. Research shows that “the uninsured tend to be disproportionately poor, young, and from racial and/or ethnic minority groups.”²⁸ Inadequate insurance limits access to basic care, not to mention the specialists needed for patients with a rare disease.

Even for those with insurance, people in poverty have difficulty when experiencing the American medical system. Missed appointments are common for poorer Americans due to logistical challenges such as working hourly jobs with limited leave, securing reliable transportation, and finding childcare. The rise of telehealth during the COVID-19 pandemic poses a promising solution to some of these logistical problems, although some medical care for rare disease patients cannot be completed virtually.

Nevertheless, research shows that differences in socioeconomic status alone do not explain America’s racial health gap.²⁹ At every level of education and income, African Americans have lower life expectancy at age 25 than whites. The life expectancy of Native Americans is even lower.³⁰ To close the gap, noneconomic factors need to be addressed.

A communication barrier between patients and their doctors also poses a challenge for patients whose first language is not English. Medical terms, especially as they relate to confusing symptoms or treatments that may relate to rare disease, can be difficult to translate. This difficulty may confuse some patients, making them increasingly reluctant to seek and receive care.³¹ Some patients may also lack insurance or be hesitant to promptly seek medical care due to their immigration status.³²

Cultural barriers play a pervasive role in the disconnect between people of color and medical care. These barriers have historical roots, including distrust of the medical system due to past mistreatment. Some of the distrust within the Black American community stems from deliberate mistreatment during the “Tuskegee Study of Untreated Syphilis,” which targeted Black men beginning in 1932 and ending in 1972.³³ Study participants were misled about the purpose of the research and were not given adequate medical treatment for their disease.

Though some of this mistreatment occurred long ago, the psychological and systemic harm remains. In a recent survey, only 54 percent of African Americans said they would definitely or probably get a COVID-19 vaccine when available to them, compared to 74 percent for white Americans.³⁴ Latinos, too, are often hesitant.³⁵ Indeed, communities of color living with rare disease may be unable or unwilling to recognize their symptoms, seek care when it is needed, and trust medical professionals to diagnose them accurately and reliably.

The medical community has begun to acknowledge historical racism. Several medical associations have formally apologized for their actions, and the American Medical Association (AMA) has called racism a public health crisis.³⁶ In January 2021, the American Psychiatric Association (APA) apologized for its support of structural racism.³⁷ Despite some progress, racial health disparities in America persist—and they particularly impact people of color with rare diseases. Until we recognize the past, it will be difficult to move forward.

A Nation Ready for Change

A growing number of Americans are beginning to see the systemic racism in America. High-profile police shootings of unarmed Black people have increased recognition of the injustices in all facets of our society. These tragedies have led to a growing consensus that something must be done.

Our nation's leaders have expressed the will to solve these problems. President Biden has made equity, including health equity, a cornerstone of his campaign and now his administration.³⁸ Once taking office, President Biden created the COVID-19 Health Equity Task Force.³⁹ Many Democratic and Republican lawmakers have spoken about racial disparities and discussed public policy solutions. Legislators on both sides of the aisle worked together to pass the First Step Act, which helped to reduce disparities in the criminal justice system.⁴⁰ More needs to be done to address health equity, especially for people of color with rare diseases, and now is the time to act. With this level of momentum and support, we can dig deep and change the social determinants of health—those conditions around how people live, learn, work, and play that affect various aspects of health and quality of life—in underserved communities.

Leading the Way: The Rare Disease Diversity Coalition

The Black Women's Health Imperative (BWHI) is an organization that deeply understands the health care challenges faced by people of color with rare diseases. Black, Hispanic, Asian and Pacific Islander, and Native American people with rare diseases often struggle with obtaining a diagnosis and adequate treatment and care. These barriers are even more pronounced and widespread among people of color, whose lack of access to health care, low representation in research and trials, and other challenges can end up causing serious health consequences. The COVID-19 pandemic has highlighted longstanding health disparities and has renewed focus on racial injustice in the United States. These conditions and the opportunities to be a catalyst for change inspired a motivated group of patient advocates and health policy experts to form the Rare Disease Diversity Coalition (RDDC) to work toward progress on rare diseases and health equity and to serve as a bridge between the two movements.

In May 2020, BWHI launched RDDC—a first-of-its-kind coalition, with the goal to address the extraordinary challenges facing people of color with rare disease. RDDC formed to work collaboratively toward an actionable path forward on both rare diseases and health equity. RDDC develops its plans through evidence-based research, representation, advocacy, and transparent acknowledgment of the challenges we face. RDDC draws strength from the coalitions that led on the ODA—which resulted in the development of NORD—as well as inspiration and a lesson that, together, we can drive progress on both rare diseases and health equity.

RDDC brings together rare disease experts, patient organizations, health and diversity advocates, and industry leaders with a deep knowledge of the medical, industry, regulatory, and cultural challenges facing people of color with rare disease. RDDC is poised to achieve action in the years to come by:

- Reducing racial disparities in the rare disease community;
- Identifying and advocating for evidence-based solutions to alleviate the disproportionate burden of rare diseases on communities of color; and
- Helping to achieve greater equality within the rare disease community.

The Coalition believes firmly in collaboration and leveraging input of stakeholders, including rare disease patients and families, who experience daily the struggles of these issues; patient organizations, who 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 changes in government policy that lead to progress.

To tap into the collective expertise and experiences of various stakeholders, RDDC initially convened workgroups to study and discuss needed progress in four key areas for people of color living with rare diseases.

1. Correcting delays in diagnosis and treatment
2. Improving patient and provider education and engagement
3. Improving the state of research and clinical trials
4. Advocating for government policy

How Recommendations Formed

The RDDC’s Steering Committee undertook a 6-month study to assess the most pressing challenges faced by people of color with rare disease and identify potential solutions. Based on a review of existing literature, research, working group analysis, and input from patients and the Steering Committee, RDDC identified more than 75 recommendations, all included in the appendix, that cut across the four key areas of focus. After identifying these recommendations, eleven were prioritized. These recommendations directly address many of the social determinants of health with a focused eye toward improving health and reducing longstanding disparities in health care for people of color with rare diseases.

These recommendations form a preliminary roadmap to guide the Coalition’s 2021 advocacy work toward specific actionable efforts that will result in progress for people of color living with rare diseases. It also sets the stage for future efforts to build a more equitable health system for people of color with rare diseases.

Top-Level Recommendations

Reducing Delays in Diagnosis and Improving Treatment and Improving Patient and Provider Education and Engagement	
<ul style="list-style-type: none"> ▶ Educate, equip, and engage health care providers to deliver culturally appropriate care to people of color living with rare diseases. ▶ Exponentially increase the number of people from marginalized groups pursuing professions in medicine and health care. ▶ Improve and increase trust in the health care system for people of color living with rare diseases. ▶ Increase engagement from communities of color in patient advocacy groups. ▶ Increase medical literacy and patient confidence among rare disease patients and caregivers of color. 	
Improving the State of Research and Clinical Trials	Advocating for Government Policy
<ul style="list-style-type: none"> ▶ Address pipeline and systemic issues that inhibit communities of color from participating in research and clinical trials by educating researchers and providers. ▶ Increase the number of rare disease researchers and medical students that come from communities of color. ▶ Increase the participation of communities of color in research and clinical trials by increasing regulatory flexibility. 	<ul style="list-style-type: none"> ▶ Increase research and treatment innovations for minority rare disease populations by passing flagship legislation to address disparities in rare disease access and care. ▶ Leverage telehealth to improve access to care and treatment for people of color living with rare diseases. ▶ Educate the administration, federal agencies, and bipartisan Congressional leaders about the importance of addressing racial disparities in the rare disease community and urge targeted action consistent with new health equity initiatives.

These recommendations are only the beginning of RDDC’s work. For each recommendation, RDDC has outlined concrete steps to make meaningful progress in 2021. All recommendations and specific action plans are set forth in [Appendix #1](#).

Working toward these recommendations will require the commitment and support of industry leaders, issue experts, health care professionals, and patients. Interested stakeholders are invited to join RDDC in this important work. With widespread support and commitment, we can take steps together toward reducing the disproportionate burden of rare diseases on communities of color.



A Path Forward—Change Is Possible

As we considered RDDC’s mission and our priorities on health equity and rare disease, the opportunity to make progress was obvious. The good work done by the coalition that pushed to develop the ODA (led by NORD) serves as a great example of how the coalition we envision can change the system effectively—to incentivize R&D in areas that were otherwise “orphaned.” This time, the mission is to continue to improve the system for patients with rare diseases and to address health disparities for people of color with rare disease.

Then, as we see now, a certain portion of the population was being left behind. Also then as now, commercial and market incentives were not sufficient to result in the developments society needed. However, a small, dedicated group of patients, advocates, and experts was able to make positive changes.

Today, RDDC, along with its allies, will begin persuading government to take basic steps for its people. Today, RDDC calls for a crusade to eliminate health disparities, especially in the realm of rare diseases, where the disparities have such devastating effects. RDDC calls for legislative and administrative solutions, for updates to existing policies, and for new policies.

This opportunity comes at a time in which many in our society support efforts to finally address racial equity, including health disparities, effectively and systemically. The time is now, with the Biden Administration and Congress identifying racial and health equity as priorities. RDDC is pleased that BWHI has engaged with policymakers from the Biden Administration and Congress and has generated excitement to work with our Coalition to make progress for the health equity and rare disease movements.

RDDC looks forward to working with you to dig deeper into these recommendations and turn them into action, drawing on the historic work of the coalitions that developed the original ODA policy fixes. We will explore the extent to which these policies may be improved to address health disparities in rare diseases or work as a playbook for separate health equity policy development. Our policy workgroup will explore how other policy considerations may be adjusted to include our Coalition’s recommendations to make progress in health disparities and rare disease policy.

Reducing Disparities Through Administrative Action

Federal health-related agencies should declare in government policies the stated objective to eliminate health disparities. The respective agencies should implement specific policies to effect real change. For example, the FDA needs to continue to develop policy to encourage diversity in clinical trials. RDDC will use the public comment process and other avenues to help push administrative agencies to adopt concrete policies to reduce health disparities.

Reducing Disparities Through Legislation

New laws must be enacted and old laws updated to eliminate health disparities. The ODA has helped the rare disease community broadly, but updates should be made to further help the community and people of color with rare diseases who have often been left behind. New and enhanced provisions need to be designed to encourage making every aspect of the health care system do its part to eliminate health disparities. The tactics used in the original ODA to address market failures with orphan drugs can be used effectively today to address health disparities.

The 21st Century Cures Act also needs to be updated to address health disparities. The current draft of Cures 2.0, a follow-up to 21st Century Cures, helps address health disparities by including policies on health literacy and diversity in clinical trials.⁴¹ However, more needs to be done to ensure and protect rare disease patients' access to high quality innovations—starting with every aspect of defining “value”. Cures 2.0 needs to make sure that the innovations it propels in gene therapy, diagnostics, and other cutting-edge areas help those who need help the most. Many components of the 21st Century Cures Act need to be updated to incentivize the development of treatments that will decrease health disparities.

Policies also must be passed to ensure that the rare disease community and people of color with rare diseases can access new treatments once they are approved. Numerous socioeconomic status and racial hurdles prevent these patients from accessing the care they need. For example, policies should be passed to prevent insurers from using prior authorization requirements and step therapy programs to prevent patients from getting the care they need in a timely manner. Lastly, RDDC supports the passage of the legislation to address disparities in rare disease access and care in 2021.

Conclusion

People of color with rare diseases face immense challenges. Due to health disparities in this country, people of color with rare diseases are in a dire situation. Thankfully, the situation is not without hope. We know what can be done. We know how passionate advocates can make a difference. Policy changes can incentivize great progress. We know how to begin to reduce racial health disparities and help these patients now.

RDDC's 11 key recommendations are just the beginning. We look forward to a broad array of suggestions on working together on strategic policy aims. Other recommendations, literature, and resources are being developed and will be available on the RDDC's webpage. We invite you to join us and be part of the solution, and we welcome your suggestions.

Acknowledgments

About BWHI

For 35 years, BWHI has been dedicated solely to improving the health and wellness of Black women and girls physically, emotionally, and financially. BWHI advances and promotes Black women's health through three focus areas: wellness programs; policy and advocacy; and research translation.

BWHI launched RDDC by convening industry, advocacy, and community leaders and by involving patient and health care guests in the initial Steering Committee and working group meetings. RDDC extends great thanks to those who have been involved since our launch; without the support of the organizations and individuals listed below, we would not be where we are today.

Steering Committee

As of February 2021, RDDC is proud to have these esteemed members of our Steering Committee:

Linda Goler Blount, MPH, Black Women's Health Imperative (BWHI)

**Steering Committee Chair*

Rev. Anthony Brownlow, M.Div., MBA, Alfred Street Baptist Church

Tammy Boyd, JD, MPH, Black Women's Health Imperative

John Burns, Burns Brothers

Shonta Chambers, MSW, Patient Advocate Foundation

Juliet Choi, JD, Asian & Pacific Islander American Health Forum (APIAHF)

Garfield Clunie, MD, National Medical Association

Donna Cryer, JD, Global Liver Institute

Debbie Drell, National Organization for Rare Disorders (NORD)

Eve Dryer, Travers Therapeutics

Beverly Francis-Gibson, MA, Sickle Cell Disease Association of America

Millicent Gorham, MBA, National Black Nurses Association

Kimberly Haugstad, MBA, ACTION Partners

Amy Hinojosa, MANA National

Julia Jenkins, MA, EveryLife Foundation

Ashley John, MS, Biotechnology Innovation Organization

Lauren Lee, MA, NephCure

Aletha Maybank, MD, MPH, American Medical Association

Cassandra McCullough, MBA, Association of Black Cardiologists

Michele Oshman, Biotechnology Innovation Organization (BIO)

Brian Thompson, MD, Association of American Indian Physicians

Tamar Thompson, MS, Alexion Pharmaceuticals, Inc.

Courtney Pieczynski Keplinger, MBA, Vertex Pharmaceuticals

Pamela Price, RN, Balm In Gilead

Elena Rios, MD, National Hispanic Medical Association

Christian Rubio, MA, MBA, Global Genes

Kim Smith-Whitley, MD, Children's Hospital of Philadelphia

Saira Sultan, JD, PCORI Advisory Panel on Rare Disease

Marshall Summar, MD, Children's National Hospital

Rev. Matthew L. Watley, M.Div., Kingdom Fellowship AME Church

Laura Weidner, JD, Epilepsy Foundation

C. Grace Whiting, JD, The National Alliance For Caregiving

Yousra Yusuf, MPH, South Asian Public Health Association

Industry Sponsors

To date, the Coalition has received the valued support of industry sponsors including founding sponsor Travere Therapeutics as well as Vertex, Amgen, Aurinia, Alnylam, Alexion, GBT, Boehringer Ingelheim, and Syros Pharmaceuticals. Additionally, RDDC has received informative and vital support from guest attendees, with organizations including the National Institutes of Health (NIH), Signify Health, Hemophilia Federation of America, the Ikari Foundation, Primary Care Collaborative, Alston & Bird, and the Haystack Project. Invaluable support from patients and patient advocates has informed our working groups' research and subsequent policy recommendations.

Anchored by the expertise of this Steering Committee and supported by a growing number of sponsors and Coalition members, RDDC will continue to advocate for solutions to alleviate the disproportionate burden of rare diseases on communities of color. **Thank you to all who have been involved.**

How to Get Involved

The year 2021 is about making actionable progress for people of color with rare diseases, focused on 11 core recommendations. RDDC believes collaboration is essential and welcomes the involvement of new committed, knowledgeable stakeholders from across the health care space.

Coalition Membership

RDDC membership is open to rare disease-, diversity-, and health care-focused organizations and companies who are invested in the Coalition's mission of reducing racial disparities in the rare disease community.

Please contact us if you are interested in becoming a member: <https://www.RareDiseaseDiversity.org/>.

Sponsorship and Industry Support

Support from sponsors and health care industry leaders enables RDDC to take meaningful steps toward change for affected communities. Sponsorships support a wide range of initiatives, including:

- Providing grants for rare disease initiatives endorsed by the Steering Committee that build the capacity for communities of color to address health disparities in policy and research;
- Developing educational materials to assist health care communities, inspire media, and engage the broader public; and
- Convening webinars and policy briefings.

RDDC welcomes additional support. Please contact us if you are interested in sponsoring our work. <https://www.RareDiseaseDiversity.org/>.

2021 Schedule of Meetings

Full Coalition meetings will take place every other month beginning in February 2021. These meetings provide members and stakeholders the opportunity to align on progress and immediate items. All RDDC members are welcome, as are guest invitees, speakers, and rare disease patients or patient advocates.

Contact RDDC

To learn more about the Rare Disease Diversity Coalition, please visit our website at <https://www.RareDiseaseDiversity.org/>.

You can also follow our updates on Twitter and Instagram @BlkWomensHealth.

<https://twitter.com/blkwomenshealth>

<https://www.instagram.com/blkwomenshealth/>

Appendix 1: 2021 Actionable Roadmap

Key RDDC recommendations and 2021 planned action steps are as follows.

Key Areas: REDUCING DELAYS IN DIAGNOSIS AND IMPROVING TREATMENT AND IMPROVING PATIENT AND PROVIDER EDUCATION AND ENGAGEMENT

Educate, equip, and engage health care providers to deliver culturally appropriate care to people of color (“POC”) living with rare diseases. This includes addressing the cultural awareness gap and apathy and ultimately impact an array of audiences, including providers, patients, caregivers, government leaders, medical institutions, and medical societies. Actions in development are to:

- Fund cultural awareness and bias training (including a pilot) for health care providers
- Implement continuing medical education (CME) in partnership with one or more medical associations or medical centers featuring underserved community engagement and interaction
- Develop a national directory of other cultural competency training programs to serve as an ongoing provider resource
- Improve medical school curriculum to include greater exposure to rare diseases

Exponentially increase the number of people from marginalized groups pursuing professions in medicine and health care. This will have indirect benefits for POC living with a rare disease. Efforts in development are to:

- Partner with the Association of American Medical Colleges to increase diversity in the pipeline at medical schools
- Work with the Association of American Medical Colleges (AAMC) to establish a list of specific recommendations to increase diverse enrollment for medical schools
- Launch an educational campaign targeted at medical schools, to hire more minority groups to their faculty

Improve and increase trust in the health care system for POC living with rare disease families.

Building trust takes time, effort and intention and the investment results in stronger overall health outcomes. RDDC efforts in development are to:

- Develop a diversity “best practices” document for the health care industry in collaboration with American Medical Association (AMA), National Medical Association (NMA), National Hispanic Medical Association (NHMA), and BIO, among others which includes best-in-class recommendations and education for members on how to develop culturally sensitive and inclusive education and marketing materials
- Utilize the “best practices” document to further promote diversity in public relations and marketing vendors for industry and organizations
- Facilitate an “industry commitment” to inclusion efforts which contains a pledge to translate materials into lay language for patients at varying educational levels and into Spanish
- Host a mentorship program for minority students

Increase engagement from communities of color in patient advocacy groups. Efforts in development are to:

- Develop a guidance document of measurable practices for patient advocacy groups to aid them in better supporting diverse rare disease patients and caregivers of color. This guidance document will address the following areas:
 - ▶ **Governance:** Ensure boards and senior staff are diverse enough to foster disparate perspectives in programming and practices
 - ▶ **Events:** Develop guidance for offering conference scholarships to ensure that families from all socio-economic groups can attend, planning committees are diverse, weekend conferences are offered for caregivers who cannot afford to take off from work, childcare is provided for families at conferences, and tailored programming is provided for Spanish-speaking attendees
 - ▶ **Communications:** Ensure communication channels such as website and social media profiles are diverse; provide bilingual or multilingual patient materials
 - ▶ **Collaborations:** Develop guidance for diversifying scientific advisory committee, create peer mentor programs to connect newly diagnosed patients with families like them, and develop community organization partnerships that specifically focus on underserved groups

Increase medical literacy and patient confidence among rare disease patients and caregivers of color. Knowledge can empower rare disease patients and caregivers of color to become active partners in their health and medical care experience. Efforts in development will equip patients to better navigate the health care system moving forward and include:

- Partnering with patient advocacy groups to conduct an educational campaign to empower diverse patients and their networks. This includes delivering online and in-person programming and collateral materials that provide guidance in areas such as:
 - ▶ The “ABCs of your newly diagnosed disease”;
 - ▶ Selecting the right care advocate;
 - ▶ Making the most of medical appointments; and
 - ▶ Speaking up or filing complaints in scenarios where treated unfairly.

Key Area: IMPROVING THE STATE OF RESEARCH AND CLINICAL TRIALS

Address pipeline and systemic issues that inhibit communities of color from participating in research and clinical trials by educating researchers and providers. Efforts in development are to:

- Provide funding to a fellow to identify best practices in rare disease research and clinical trial studies that had a large number of participants from communities of color
- Develop a summary guidance document of best practices, with target audiences including the broader medical community and POC
- Utilize identified best practices to educate researchers and providers with the know-how to engage with communities of color in order to reach, educate, build relationships, and ultimately increase participation of enroll large numbers of POC in research and clinical trials

Increase the participation of communities of color in research and clinical trials by increasing regulatory flexibility. This will create synergies with FDA given mutual interests in innovation, collaboration and in developing treatment therapies for rare diseases. To accomplish this effort, RDDC will engage regulators in an ongoing dialogue, providing opportunities for the rare disease community to inform regulatory reviews and provide feedback on how to improve clinical trials for POC.

Efforts in development are to:

- Create a Rare Disease & Diversity Center of Excellence focused on racial and ethnic minorities at the FDA
- Work with the FDA to train investigators and formulate data that can be useful to the agency in improving participation of POC in clinical trials

Increase the number of rare disease researchers and medical students who come from communities of color. Programs and initiatives will positively impact representation as well as prioritize research monies that focus on rare diseases that disproportionately affect communities of color. Efforts in development are to:

- Create a rare disease fellowship focused on communities of color
- Establish a loan repayment program (LRP) for medical students in communities of color to specialize in rare disease specialties (e.g., genetics)
- Increase mentorship programs for communities of color in rare disease (e.g., R25 training program with the NIH)

Key Area: ADVOCATING FOR GOVERNMENT REGULATION, LEGISLATION, AND POLICY

Increase research and treatment innovations for minority rare disease populations by passing flagship legislation to address disparities in rare disease access and care. This important legislation would require directors at the NIH, Centers for Disease Control and Prevention (CDC), the Health Resources and Service Administration (HRSA), HHS, and the Indian Health Service (IHS) to take actions related to rare diseases in minority populations. Efforts in development are to:

- Require the director of the NIH to expand research with respect to rare diseases in minority populations
- Expand the Minority Access to Research Careers program
- Direct the NIH and CDC to submit a comprehensive federal plan to Congress to address rare diseases in minority patients and a comprehensive list of rare conditions that impact communities of color
- Require the director of the CDC to conduct research related to POC with rare diseases and require the director of HRSA to educate health professionals on the causes and effects of rare diseases in minority populations
- Direct the NIH & CDC to submit a report on federal research and public health activities to Congress with respect to rare diseases in minority populations.

Leverage telehealth to improve access to care and treatment for patients of color living with rare diseases. Advocacy efforts will support continued telehealth access for rare disease diverse patients, both during and after COVID-19. Key challenges in telehealth such as technology barriers, rules, and restrictions that limit the permanent availability of telehealth options for patients are key areas of focus. Efforts in development are to:

- Support efforts to make the telehealth waiver permanent and address telehealth licensing barriers
- Advocate for the Evaluating Disparities and Outcomes of Telehealth Act which would study the effects of changes to telehealth under the Medicare and Medicaid programs during the COVID-19 emergency, including changes affecting minority populations
- Coordinate with advocacy partners to evaluate other legislative opportunities related to expanding telehealth options

Educate the Biden Administration, federal agencies, and bipartisan congressional leaders about the importance of addressing racial disparities in the rare disease community and urge targeted action consistent with new health equity initiatives. Efforts in development are to:

- Create educational materials to share with government officials
- Conduct proactive outreach on a bipartisan basis to identify and build support for new legislative and regulatory solutions.

**Numerous other suggestions and ideas were captured in the RDDC Working Groups during 2020 that have future potential for action. The above RDDC recommendations and action plans for 2021 are viewed as a positive first step to making real progress in both 2021 and in the years to come.*

References

- 1 Genetic and Rare Diseases Information Center, NIH. "FAQs about Rare Diseases." U.S. Department of Health and Human Services, 2021, rarediseases.info.nih.gov/diseases/pages/31/faqs-about-rare-diseases.
- 2 Novorol, Claire. "The Challenges of Combating Rare Diseases—And Five Innovations Making a Real Difference." *Forbes*. 28 Feb. 2020, www.forbes.com/sites/clairenovorol/2020/02/28/the-challenges-of-combating-rare-diseases-and-five-innovations-making-a-real-difference/?sh=7ce132a35a58.
- 3 Global Genes. "RARE Facts." Global Genes, 16 Dec. 2020, globalgenes.org/rare-facts/.
- 4 United States Congress. *Hearing before Subcommittee on Health and Environment of the House Committee on Interstate and Foreign Commerce on Drug Regulation Reform: Oversight on Orphan Drugs*. 96th Congress, 2nd session (1980) (statement of Abbey Meyers, Tourette Syndrome Association, page 20). <https://babel.hathitrust.org/cgi/pt?id=mdp.39015004895994&view=1up&seq=24>.
- 5 Congressional Research Service. 2001, July 25. Orphan Drug Act: Background and Proposed Legislation in the 107th Congress (CRS Report No. RS20971). https://www.everycrsreport.com/files/20010725_RS20971_b20ead0a2eb41ebdc6388f88d2e881fd7f36acae.pdf.
- 6 Significant Drugs of Limited Commercial Value, Interagency Task Force report to the Secretary of Health, Education, and Welfare. June 29, 1979. https://rarediseases.info.nih.gov/files/1979_interagency_task_force_report_on_significant_drugs_of_limited_clinical_value.pdf.
- 7 Asbury CH. The Orphan Drug Act: The First 7 Years. *JAMA*. 1991;265(7):893-897. doi:10.1001/jama.1991.03460070075046.
- 8 Koichi Mikami, Orphans in the Market: The History of Orphan Drug Policy, Social History of Medicine, Volume 32, Issue 3, August 2019, Pages 609-630, <https://doi.org/10.1093/shm/hkx098>.
- 9 Orphan Drug Act of 1983. Pub L. No. 97-414, 96 Stat. 2049.
- 10 NORD. "What Is the Orphan Drug Act?" <https://rarediseases.org/>, NORD, 2019, rarediseases.org/wp-content/uploads/2019/10/NRD-1159-RI-WhatIsTheODA_2019_FNL-1.pdf.
- 11 Packer, Milton. Has Drug Development for Rare Diseases Reached an Extreme? *MedpageToday*, 22 Aug. 2018, www.medpagetoday.com/blogs/revolutionand revelation/74687.
- 12 FDA. "Search Orphan Drug Designations and Approvals." [Accessdata.fda.gov](https://accessdata.fda.gov), HHS, 16 Feb. 2021, www.accessdata.fda.gov/scripts/opdlisting/opd/index.cfm.
- 13 Thorat, C., et al. "What the Orphan Drug Act Has Done Lately for Children With Rare Diseases: A 10-Year Analysis." *PEDIATRICS*, vol. 129, no. 3, 27 Mar. 2012, pp. 516-521., doi:10.1542/peds.2011-1798.
- 14 Office of the Commissioner. "21st Century Cures Act." FDA, 31 Jan. 2020, www.fda.gov/regulatory-information/selected-amendments-fdc-act/21st-century-cures-act.
- 15 APHL. "Newborn Screening Status for All Disorders." *NewSTEPS*, 2021, www.newsteps.org/resources/data-visualizations/newborn-screening-status-all-disorders.
- 16 Office of the Commissioner. "Fast Track, Breakthrough Therapy, Accelerated Approval, Priority Review." U.S. Food and Drug Administration, HHS, 23 Feb. 2018, www.fda.gov/patients/learn-about-drug-and-device-approvals/fast-track-breakthrough-therapy-accelerated-approval-priority-review.
- 17 Fassbender, Melissa. "Natural History Studies 'Essential' for Rare Disease Drug Development." *Outsourcing Pharma*, William Reed Business Media Ltd., 17 July 2019, www.outsourcing-pharma.com/Article/2019/07/17/Natural-history-studies-essential-for-rare-disease-drug-development.
- 18 IQVIA Institute. "Orphan Drugs in the United States." www.iqvia.com, 2018, www.iqvia.com/-/media/iqvia/pdfs/institute-reports/orphan-drugs-in-the-united-states-exclusivity-pricing-and-treated-populations.pdf?_=1548445322680.
- 19 Global Genes. "Rare Disease Impact Report: Insights from Patients and the Medical Community." *Globalgenes.org*, Shire, 2013, globalgenes.org/wp-content/uploads/2013/04/ShireReport-1.pdf.
- 20 Taylor, Karen. "Why Improving Inclusion and Diversity in Clinical Trials Should Be a Research Priority." *Deloitte United States*, 4 Sept. 2019, www2.deloitte.com/us/en/blog/health-care-blog/2019/why-improving-inclusion-and-diversity-in-clinical-trials-should-be-a-research-priority.html.
- 21 Pfizer. "Health Disparities Among African-Americans." *Your Health*, Pfizer, 9 Sept. 2020, www.pfizer.com/news/hot-topics/health_disparities_among_african_americans.
- 22 Black Health Matters. "Rare Diseases and African Americans." *BlackHealthMatters.com*, 6 Mar. 2020, blackhealthmatters.com/health-conditions-hub/hattr-amylodosis/rare-diseases-and-african-americans/.
- 23 Hall, William J., et al. "Implicit Racial/Ethnic Bias Among Health Care Professionals and Its Influence on Health Care Outcomes: A Systematic Review." *American Journal of Public Health*, vol. 105, no. 12, Dec. 2015, pp. e60-e76., doi:10.2105/ajph.2015.302903a.
- 24 ODPHP. "Social Determinants of Health." *Healthy People 2020*, HHS, 8 Oct. 2020, www.healthypeople.gov/2020/topics-objectives/topic/social-determinants-of-health.
- 25 Wang, Jian, and Liuna Geng. "Effects of Socioeconomic Status on Physical and Psychological Health: Lifestyle as a Mediator." *International journal of environmental research and public health* vol. 16,2 281. 20 Jan. 2019, doi:10.3390/ijerph16020281.
- 26 Bhutta, Neil. "Disparities in Wealth by Race and Ethnicity in the 2019 Survey of Consumer Finances." *The Fed*, Board of Governors of the Federal Reserve System, 28 Sept. 2020, www.federalreserve.gov/econres/notes/feds-notes/disparities-in-wealth-by-race-and-ethnicity-in-the-2019-survey-of-consumer-finances-20200928.htm.
- 27 Benfer, Emily A. "Contaminated Childhood: The Chronic Lead Poisoning of Low-Income Children and Communities of Color in the United States." *Health Affairs*, Project Hope, 8 Aug. 2017, www.healthaffairs.org/doi/10.1377/hblog20170808.061398/full/.
- 28 Riley, Wayne J. "Health disparities: gaps in access, quality and affordability of medical care." *Transactions of the American Clinical and Climatological Association* vol. 123 (2012): 167-72; discussion 172-4.
- 29 Williams, David R et al. "Racism and Health: Evidence and Needed Research." *Annual review of public health* vol. 40 (2019): 105-125. doi:10.1146/annurev-publhealth-040218-043750.
- 30 Indian Health Service. 2019. Fact Sheet: Disparities. <https://www.ihs.gov/newsroom/factsheets/disparities/>.
- 31 Al Shamsi, Hilal et al. "Implications of Language Barriers for Healthcare: A Systematic Review." *Oman medical journal* vol. 35,2 e122. 30 Apr. 2020, doi:10.5001/omj.2020.40.

- 32 Artiga, Samantha, and Maria Diaz. "Health Coverage and Care of Undocumented Immigrants." KFF, Kaiser, 15 July 2019, www.kff.org/racial-equity-and-health-policy/issue-brief/health-coverage-and-care-of-undocumented-immigrants/.
- 33 NCHHSTP. "Tuskegee Study: Timeline." CDC.gov, CDC, 2 Mar. 2020, www.cdc.gov/tuskegee/timeline.htm.
- 34 Gramlich, John, and Cary Funk. Black Americans Face Higher COVID-19 Risks, Are More Hesitant to Trust Medical Scientists, Get Vaccinated. Pew Research Center, 4 June 2020, www.pewresearch.org/fact-tank/2020/06/04/black-americans-face-higher-covid-19-risks-are-more-hesitant-to-trust-medical-scientists-get-vaccinated/.
- 35 National Public Radio. February 1, 2021. Many Latinos Are Hesitant To Get A COVID-19 Vaccine <https://www.npr.org/2021/02/01/962905232/many-latinos-are-hesitant-to-get-a-covid-19-vaccine>.
- 36 American Medical Association. November 16, 2020. AMA: Racism Is a Threat to Public Health. <https://www.ama-assn.org/delivering-care/health-equity/ama-racism-threat-public-health>.
- 37 American Psychiatric Association. "APA's Apology to Black, Indigenous and People of Color for Its Support of Structural Racism in Psychiatry." www.psychiatry.org, American Psychiatric Association, 18 Jan. 2021, www.psychiatry.org/newsroom/apa-apology-for-its-support-of-structural-racism-in-psychiatry.
- 38 Biden Harris 2020. "The Biden Plan to Build Back Better by Advancing Racial Equity Across the American Economy." Joe Biden for President: Official Campaign Website, 18 Oct. 2020, joebiden.com/racial-economic-equity/.
- 39 Biden, Joseph R. "Executive Order on Ensuring an Equitable Pandemic Response and Recovery." The White House, 21 Jan. 2021, www.whitehouse.gov/briefing-room/presidential-actions/2021/01/21/executive-order-ensuring-an-equitable-pandemic-response-and-recovery/.
- 40 GovTrack. "S. 756: FIRST Step Act -- Senate Vote #271." GovTrack.us, 18 Dec. 2018, www.govtrack.us/congress/votes/115-2018/s271.
- 41 DeGette, Diana, and Fred Upton. "21st CENTURY CURES 2.0 DRAFT." [Degette.house.gov](http://degette.house.gov), 27 Apr. 2020, degette.house.gov/sites/degette.house.gov/files/Cures%202.0%20Concept%20Paper_0.pdf.



THE RARE DISEASE DIVERSITY COALITION