REGULAR ARTICLE

What Rare Disease Patient Advocacy Groups Are Doing to Mitigate the Effects of Disparities

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Compared with chronic diseases affecting larger populations, rare disease (RD) patients experience great inequities in diagnosis, care, and research. Within RDs, health disparities compound these inequities, as marginalized communities experience additional barriers in accessing clinical care and are often underrepresented from participation in research and clinical trials. For almost 40 years, the National Organization for Rare Disorders (NORD), a RD umbrella organization with over 300 nonprofit organizational members, has led efforts to understand and address inequities for the RD community through innovative research, programming, and collaboration with patients, caregivers, practitioners, and external stakeholders. The beginning of the COVID-19 pandemic in 2020 brought to light longstanding disparities and discrimination for marginalized communities as well as pivotal racial justice movements. These events spurred many RD nonprofit organizations' interest in increasing outreach and engagement with minoritized communities within RDs and diversifying their organization internally. Building on the increased interest in diversity, equity, and inclusion (DEI), NORD has focused on collecting case studies from within NORD and its member organizations to capture current efforts to improve DEI within the RD ecosystem. One way clinicians can work to mitigate the effects of disparities is to collaborate with RD patient organizations; this article provides a means by which clinicians and researchers can understand some of the challenges RD nonprofit organizations face in bridging disparities and learn about solutions to supporting marginalized patients within their communities. Clinicians are encouraged to join NORD in our policy efforts advocating to ensure patient access to health care providers practicing in a different state vis-à-vis telehealth.

INTRODUCTION TO NORD

For those active in the pulmonary hypertension (PH) advocacy space, the origin story of the Pulmonary Hypertension Association may be familiar: in the early 1990s, 3 patients and a nurse caregiver met around a kitchen table in Florida with lofty goals of support, education, and cures.[1] What may not be known is that these patients found each other by writing letters to the National Organization for Rare Disorders (NORD). National Organization for Rare Disorders staff not only connected them with one another for this historic first meeting of PH patients but provided support as their fledgling nonprofit grew: "For most of the early and mid-1990's (sic) the organization was a kitchen table operation regularly seeking advice on organizational and management issues from NORD."[2(p2)]

National Organization for Rare Disorders' history is rich with stories of helping patients and caregivers connect and advocate as central, equal partners in research and drug development. Often alone, geographically isolated, misunderstood by their family and friends, and afraid after delayed diagnoses led to advanced disease progression, people living with rare disease (RD) face inequities in access to care, research participation, and lack of treatment options.

It was the lack of treatments in the late 1970s that spurred a call to action leading to the creation of an ad hoc coalition of RD leaders who sought to change the research paradigm during a time when little was being done to study RDs or develop treatments. After successfully advocating for the passage of the Orphan Drug Act of 1983 (which created financial incentives for the development of treatments for RDs), this coalition became the foundation of NORD.[3]

For almost 40 years, NORD has served as the hub of the RD community, leading efforts to drive progress, overcome inequities experienced by patients across all RDs, and advocate for the collective representation of all affected by RDs in the United States. During this COVID-19 pandemic era of heightened international awareness of health disparities, NORD has brought increased attention to engaging historically marginalized groups—such as people of color, low-income Americans, and members of the lesbian, gay, bisexual, transgender, queer or questioning, intersex, asexual, including other sexual identities (LGBTQIA+) community—

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face greater barriers to accessing and receiving health care.[4]

In this piece, we endeavor to answer the following questions:

- What are the general inequities experienced by patients across all RDs? What issues affect all patients with RDs that are not experienced by common, well-known conditions?
- Do certain RDs face greater inequities more so than others? If so, why?
- Within a RD, are there communities that experience barriers to access of diagnosis, care, research, support, information, and treatment?
- What have other RD organizations done to address inequities in their community?
- What programs, services, and resources have NORD—the leading umbrella organization in the United States—undertaken to mitigate the effects of disparities?

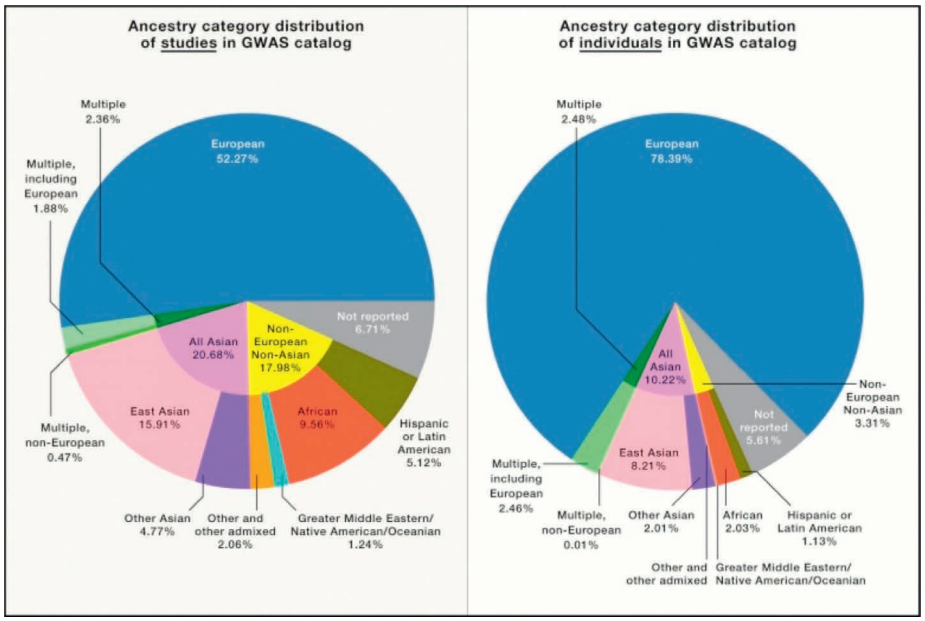

Figure 1: The missing diversity in human genetic studies. Reprinted with permission from Sirugo et al.[11]

DEFINING DISPARITIES

For the purposes of capturing the current landscape of RDs as it relates to health disparities, we define "health disparity" as a systematic, conceivably avoidable health difference according to characteristics associated with marginalization or discrimination, such as race, ethnicity, nationality, skin color, socioeconomic resources, religion, geography, gender, sexual orientation, gender identity, age, disability, illness, political, or other affiliation.[5]

Collectively, RDs are widely recognized as a public health challenge due to significant disparities, including delays and barriers to diagnosis, access to treatment, and quality management of care (eg, access to specialists).[6] It is estimated that approximately 400 million people live with a RD globally, and on average, it can take up to 5 years for people to receive an accurate diagnosis.[7,8] Though more than 7000 RDs have been identified, only 10% of these conditions have US Food and Drug Administration (FDA) approved treatments.[9] These barriers, along with an overall paucity of medical knowledge, research, and literature, leave millions of people living with RDs with inadequate resources to manage their care.

This challenging health care landscape is further magnified for people belonging to marginalized communities living with rare conditions. Eighty percent of RDs are genetic in origin, and genome-wide association studies (GWAS) play a crucial part in identifying them.[10] A 2018 analysis of GWAS revealed that, of the individuals included in studies, 78% were European, while only 10% were Asian, 2% were African, and 1% were Hispanic. All other ethnicities represented <1% of GWAS (Figure 1).[11] The implications of the lack of diversity in genomic studies can have rippling effects on communities of color seeking accurate diagnoses as well as effective treatments for their RD. This can be seen in genetic carrier screening for RDs like cystic fibrosis (CF), where a 25-mutation carrier screening panel considered to be panethnic detected close to 90% of CF carriers in white and/or Ashkenazi Jewish populations. The same panel detected only 72% of carriers in Hispanic Americans, 64% of carriers in African Americans, and 49% of carriers in Asian Americans.[12]

Disparities are not only evident in the lack of accurate diagnostic tools for diverse populations in RD but also in funding for research. While CF affects less than half the number of people as sickle cell disease (SCD) does, CF has received more than 3.5 times the amount of funding from the National Institutes of Health (NIH) and 440 times the funding from national foundations (Figure 2). Industry was also more likely to fund CF versus SCD trials (mean ± SD trials, 15.6 ± 5.3 versus 6.8 ± 1.8; $P = .001$). This difference in funding is thought to possibly correlate with a decrease in research output as well as delayed drug development for SCD. From 2008 to 2018, CF had 4 new FDA drug approvals, while SCD had 1; there were 11 novel FDA drug indications for CF, and only 2 for SCD.[13]

Recent studies in RDs have examined how the impact of social determinants of health—which include economic stability, education access and quality, health care access and quality, neighborhood and built environment, and social and community context—can impact people in different marginalized groups, including race, gender, geographic region, and socioeconomic status (SES).[14] Using US data for multiple causes of death between 1999 and 2016, authors of a 2019 study analyzed all sarcoidosis-related deaths. The highest number of sarcoidosis-related multiple-cause-of-death

Table 2. Disease-Specific NIH Funding and Combined Foundation Expenditures

Funding or expenditure	Year										Mean (SD)	*P* value
	2008	2009	2010	2011	2012	2013	2014	2015	2016	2017		
NIH funding (in millions), $												
SCD	80	63	73	65	65	70	75	75	92	105	76.3 (13.2)	.05
CF	90	86	86	79	86	78	77	80	89	91	84.2 (5.3)	
Per person affected												
SCD	889	700	811	722	722	778	833	833	1022	1167	812 (147)	<.001
CF	3000	2867	2867	2633	2867	2600	2567	2667	2967	3033	2807 (175)	
CF:SCD ratio of NIH funding per person	3.38	4.1	3.53	3.65	3.97	3.34	3.08	3.2	2.9	2.6	3.37 (0.46)	NA
Foundation expenditures (in millions), $												
SCD	10.3	9.83	9.27	8.42	7.38	7.73	8.03	9.13	11.2	10	9.14 (1.23)	<.001
CF	199	175	109	175	148	163	171	313	487	367	231 (119)	
Per person affected												
SCD	115	109	103	94	82	86	89	101	124	112	102 (13.7)	<.001
CF	6634	5823	3644	5816	4928	5443	5715	10 428	16 227	12 240	7690 (3974)	
CF:SCD ratio of foundation expenditures per person	58	53	35	62	60	63	64	103	131	109	75 (30)	NA

Abbreviations: CF, cystic fibrosis; NA, not applicable; NIH, National Institutes of Health; SCD, sickle cell disease.

Figure 2: Comparison of US federal and foundation funding of research for sickle cell disease and cystic fibrosis and factors associated with research productivity. Reprinted with permission from Farooq et al.[13]

mortality rates were found in non-Hispanic Black people and those identifying as female.[15] Similarly, it is well established that geographic location, one's zip code—even from block to block—can affect not only a person's quality of life but also their lifespan.[16] In an effort to document sarcoidosis patient-reported challenges in receiving care based on low- and high-income zip codes, authors of a study revealed that patients in both income brackets shared similar concerns in their health care management but noted that people residing in the low-income zip code communities more often reported concerns about racially biased discrimination in care as well as income bias.[17]

For certain populations, like the Amish community, geographic and technological barriers can play a role in access to health care. Culturally, many in the community choose not to use technology such as cellular phones, computers, or the Internet. Due to the founder effect, the Amish community is also disproportionately affected by certain RDs, including pyruvate kinase deficiency (PKD).[18] In September of 2019, NORD hosted an externally led Patient Focused Drug Development (EL-PFDD) meeting, sharing the experiences of patients and caregivers impacted by PKD directly with FDA regulators. While living only 300 miles away from the site of the meeting, the PKD community was unable to participate in person because they travel by horse-drawn carriages and are unable to participate in telephone and Web polling because they choose not to use the technology. To ensure their equitable access to participate in the EL-PFDD and share their voices and experiences with stakeholders, NORD staff collaborated with a leading clinician in central Pennsylvania who hosted a Patient Day so that NORD could "bring the meeting to them," explaining the reason why their perspectives are important to FDA and what they hoped to gain from collecting paper survey responses in person.[19]

Efforts like NORD's are essential to help identify gaps and barriers for marginalized communities as well as build relationships within the communities to address these gaps. Rare disease advocacy groups play a critical role in connecting people affected by RDs with up-to-date research, resources, and support services, all of which can improve their access to quality health care, treatments, and potential cures.

CASE STUDIES FROM NORD MEMBER ORGANIZATIONS

Since 2017, Angioma Alliance (AA), a patient advocacy and research organization serving those with cerebral cavernous malformation (CCM), has dedicated 25% of its annual budget to diversity, equity, and inclusion (DEI) efforts. Most recently, the organization launched an initiative to address diagnostic and treatment disparities for Black CCM (B-CCM) patients.

The Need

It is well documented that there is a disproportionate number of CCM cases in Hispanic Americans of Mexican descent. However, published studies on CCM prevalence rates in African Americans have been severely lacking, and there is a significant disparity in B-CCM patient engagement. Authors of studies suggest there should be

The Problem: Largest CCM Patient Databases in the US

Institution/Project	Total US Adult Registrants	Total Black Registrants	% Black Registrants
Angioma Alliance	914	16	1.75%
U of Chicago	512	45	8.8%
BVMC	537	2	0.4%
Mayo Clinic	282	5	1.8%
Total	**2245**	**68**	**3.0%**

Figure 3: Cerebral cavernous malformation (CCM) patient databases.

17000 Black Americans diagnosed with CCM,[20] yet in 2020, there were only 68 patients who identified as Black in the 4 major US clinical databases (Figure 3).

In 2021, AA launched Breaking Barriers for Black Health Empowerment to understand B-CCM patient experiences and develop culturally appropriate interventions to improve self-advocacy, care, and research engagement.

Angioma Alliance patient registry data pointed to clinical differences between B-CCM patients and the larger registrant cohort. Black CCM patients reported greater disability, with an average Modified Rankin Scale score of 2 in comparison with an average score of 1.3 in all other registrants. Fifty percent of Black registrants reported spinal cord lesions. Spinal cord lesions are an uncommon finding in the greater patient cohort (<15%), and hemorrhage in these lesions leads to acute, often severe, symptoms. Less dramatic CCM symptoms—mild functional neurological deficits, headache, and partial complex seizure—may not lead to diagnosis in Black patients possibly because patients may not receive appropriate referrals.[21]

The Breaking Barriers program also conducted a preliminary qualitative study of B-CCM patients using semistructured interviews which were transcribed and coded. Contrary to expectations, this cohort did not report delayed diagnosis or limited access to quality acute care. Instead, B-CCM patients universally reported a lack of aftercare referrals. This resulted in little patient education or health monitoring and nonexistent mental health or case management services. While underdiagnosis may play a role in reduced patient engagement, lack of aftercare services may have an equivalent or greater impact.

Program

The Breaking Barriers initiative has launched a 4-pronged approach to the challenge of health disparities among B-CCM patients (Figure 4). First, the program is facilitating the development of a cohesive community between known B-CCM patients through virtual connections and medical education events. Membership in a private Facebook group and Zoom support groups provide patients with access to invited CCM expert guest speakers and up-to-date disease information.

Second, the program supports aftercare through a "second look" program in which B-CCM patients are offered the opportunity to meet one-on-one with AA staff for needs assessment

Breaking Barriers Initiatives

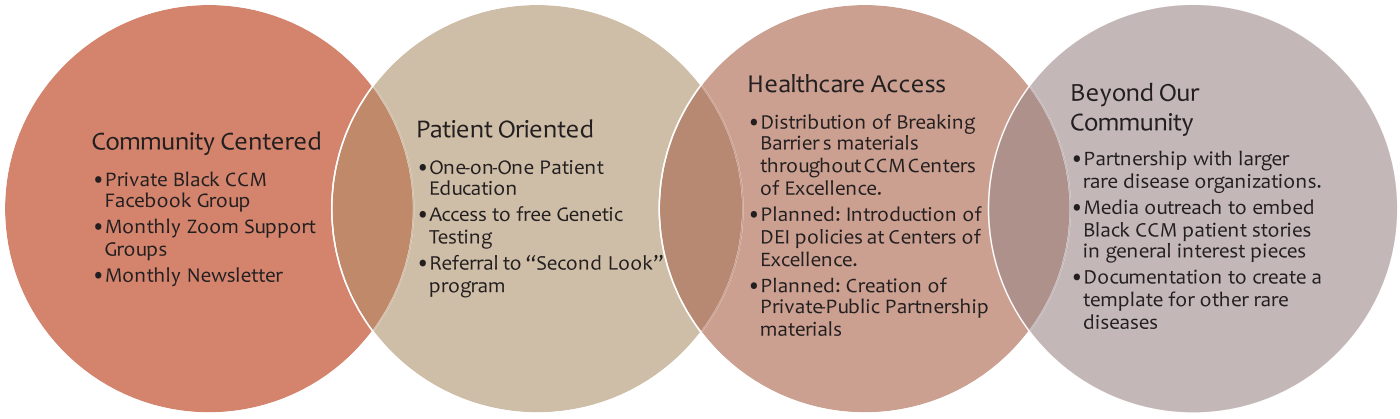

Figure 4: Breaking Barriers initiative.

and referrals to CCM experts; specific AA staff have decades of experience in the community, are trained in patient engagement/sensitivity, serve as case managers, and do not provide medical guidance; one staff member is a licensed clinical psychologist and another has a master's in public health. A third staff member was hired who is a registered nurse and is a certified case manager. Angioma Alliance also provides free genetic testing for patients where appropriate (meeting phenotype criteria) and individual patient education to bolster health self-advocacy.

Third, the program addresses disparities within clinical research and care. The CCM Health Index, an industry-sponsored patient-reported outcomes measure (PROM), required multiple rounds of recruitment through the varying phases of its development. In the initial cross sectional study phase, 323 participants were recruited, 13% of whom identified as members of racial or ethnic minorities. Subsequently, for the longitudinal study phase of development, the organization successfully advocated for patient compensation. Additionally, targeted outreach and communication strategies, like doubling email messaging to Black and Latinx patients living with CCM, were implemented to improve reach and engagement with marginalized and underrepresented patients. Angioma Alliance recruited 616 patients for the longitudinal study phase, and, as a result of these efforts, 29% of this cohort identified as members of minority groups. Angioma Alliance is now adding DEI requirements to CCM Center of Excellence criteria and is developing continuing medical education materials to assist with diagnosis and care in community hospital settings.

Challenges and Learnings

While the first year of Breaking Barriers was successful, the program faced several challenges. In response to the initiative announcement, a major donor withdrew their support. Foundation support mitigated the loss and offset the budgetary impact.

The initiative struggled with patient engagement. The B-CCM registry increased by 250% in the first year of the program (Figure 5); unfortunately, the absolute number remained small. The engagement challenge was particularly evident with male patients. As an example, semistructured interviews were conducted with 11 women, but only 2 men were willing to participate.

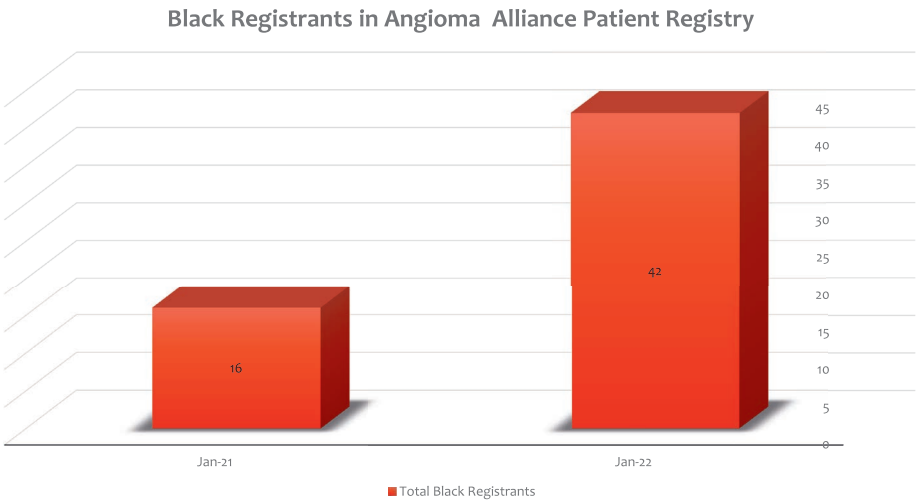

Figure 5: Growth in Black patients in the Angioma Alliance registry.

Finally, a Breaking Barriers support group discussion led to an unexpected finding. At least 5% of familial CCM cases include vascular skin lesions—deep blue nodules, punctate capillary malformations, and hyperkeratotic capillary venous malformations.[22] Support group patients requested assistance with lesion identification. An expert in CCM cutaneous lesions scoured the literature but could find no examples of these lesions on Black skin. This initiated a project to collect images within the Breaking Barriers patient cohort for a potential future publication.

Angioma Alliance's Black Health Empowerment (Breaking Barriers) program is moving beyond the B-CCM community. Breaking Barriers hired a media relations firm to identify opportunities to feature B-CCM patients in larger mainstream media stories.

Although this program may have lost a major donor, the merit of the initiative attracted support from other funders, including a multiyear grant from the Chan Zuckerberg Initiative Rare as One program, a grant program for which only 50 of the 1200 RD nonprofit organizations were selected for funding in its first 3 years of grant cycles. National Organization for Rare Disorders supports the AA and shares their work as a model to the 1200 organizations they serve.

NATIONAL ORGANIZATION FOR RARE DISORDERS' WORK IN MITIGATING THE EFFECTS OF HEALTH DISPARITIES

Supporting RD Nonprofit Capacity

Previous case studies exemplify the more advanced RD nonprofit organizations in our network. The AA has a team of 9 full-time employees (FTEs), and Sarcoidosis Research Foundation has a team of 11 FTEs.[23] However, in NORD's 2018 survey of our member organizations, 81% of our members have fewer than 5 FTEs, and the majority are managed with no FTEs (Figure 6); 1 in 5 operate with a budget of less than $25000 and nearly half with less than $200000.[24] Essentially, our survey demonstrates many RD organizations are underresourced and do not have the means to implement DEI principles within their work, despite their tremendous interest.

In addition, the leadership, staff, and board of directors of RD nonprofits do not always represent the diversity of their communities; across all nonprofits, board members are 78% white, while about 61% of Americans are white.[25] Rare disease nonprofit leadership likely reflects these trends in America: homogenously white and of a mid to upper

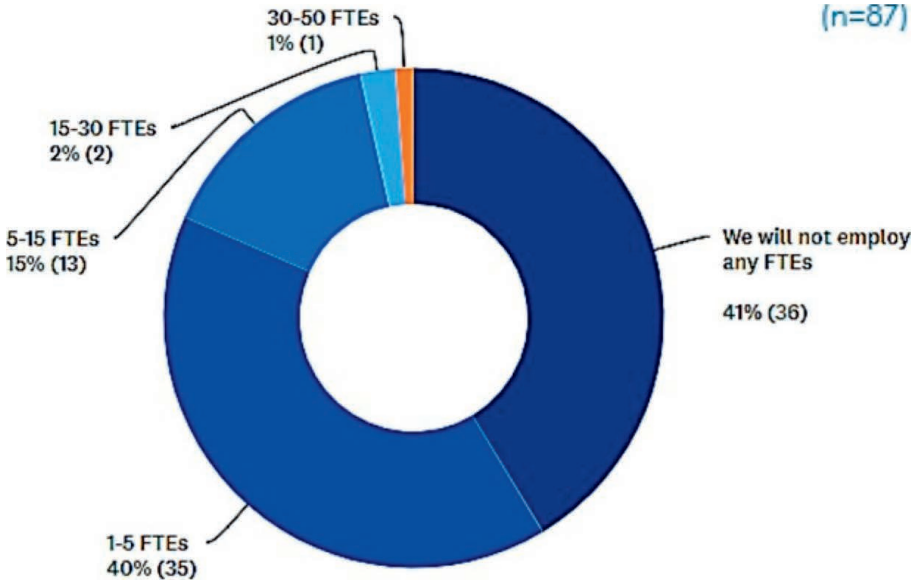

Figure 6: National Organization for Rare Disorders 2018 survey of full-time employees (FTEs).

level of SES. As a result, their outreach may fail to reach or even recognize marginalized communities impacted by their RD, and subsequently. patient engagement with their organization may not reflect the full diversity of the disease state.

National Organization for Rare Disorders is the oldest RD umbrella nonprofit in the United States—a leader in the RD patient advocacy ecosystem with a rich history of helping to establish many of its member organizations, providing best practices, education, and training. Given these close ties, NORD is well positioned as a trusted convener and seen as exemplary in assisting patient organizations to examine their activities and adopt more inclusive practices. In the fall of 2021, NORD developed a 3-part webinar series and 3-part toolkit providing leaders and staff of RD patient organizations a deeper understanding of how health disparities in marginalized populations affect the RD community as well as suggesting strategies to authentically connect with people of color, those who face language and/or literacy barriers, and those in remote or underserved areas who may lack access to online resources and the medical expertise necessary to manage their care. More than 1000 leaders of hundreds of nonprofit organizations registered and participated in the webinar series, which focused on (1) foundational knowledge of concepts around DEI and its intersectionality with RDs, (2) inclusion practices and their role in RD advocacy, and (3) the importance of diversifying nonprofit leadership and strategies to increase representation on nonprofit board of directors.

Supporting Treatment Access for Low-Income RD Patients

Commencing in 1987, NORD's patient assistance program is one of the first in the nation, providing financial support for helping low-income patients obtain lifesaving or life-sustaining medication they could not otherwise afford. These programs provide eligible individuals with financial assistance for health insurance premiums, copay costs for medical consultations, medications, diagnostic tests, and supportive therapies. Additionally, they may provide financial support for travel for clinical trials and/or consultations with disease specialists.

In 2021, NORD awarded $40698802 in patient assistance, helping 9419 patients with 265 RDs across the United States and 5 US territories. Of these patients, 959 were assisted with medical expenses not covered by health insurance and 7636 assisted with health insurance premiums and/or copay expenses. We offer travel and lodging assistance for 11 clinical trials.

When we set up a program, we look at the disease state and demographic makeup of its population—is there a higher risk or propensity for specific age, race, ethnic, or religious groups? When we identify a population being disproportionately older, we consider possible technology barriers and design resources to make enrollment in our program accessible to this community. Similarly, when we identify a population as having a higher proportion of Spanish speakers, we will develop materials in Spanish. We have several members of our team who are fluent in Spanish, as this language is the most widely spoken in the United States outside of English.[26] We want our programs to be accessible to all ethnicities in the United States, so we use Language Line, a telephone service that offers on-demand live translation of various languages into English. Thus, even if a caller is unable to speak English but fluent in say, Mandarin or Portuguese, we can just access Language Line for immediate translation.

We promote our patient assistance programs working with our members and the 1200 RD nonprofits in our network. In the future, we hope to use our new Centers of Excellence to develop new outreach programs to marginalized communities. To date, our centers are in 21 states and the District of Columbia, including largely rural areas in Alabama, Nebraska, and Oklahoma, to name a few.[27]

Our Patient Assistance Program not only supports patients directly, but our program model and learnings from our work are passed onto our member organizations when they approach us for assistance in setting up their own programs.

National Organization for Rare Disorders' Legislative Action for Bridging Disparities

Since our inception and for nearly 40 years, NORD has been the leader in advocating at the federal and state level for health policy designed to improve the lives of RD patients by increasing access to the following:

- Affordable, comprehensive health care coverage;
- New and innovative therapies to treat RDs; and
- Diagnostic tools that will enable early and accurate diagnosis.

In recent history, we have worked to support paid family and medical leave, as well as telehealth.

This includes our work in supporting the Affordable Care Act's essential health benefits and affordability provisions, advocating for Medicaid expansion at the state level, supporting patient protections to enable timely prescription drug access, and opposing Medicaid eligibility restrictions. Since 2017 alone, we have drafted and asserted our position vis-à-vis 812 policy statements, congressional and state legislature testimonies, and cosigned letters on RD health care access and research.[28]

Working within various coalitions with large and small nonprofits, we have spearheaded the RD response to policy by bringing hundreds of organizations together to elevate and integrate the RD community's needs into broader patient advocacy efforts within our health care system. At the state level, our Project RDAC is working to establish robust Rare Disease Advisory Councils (RDAC) in all 50 states to analyze the needs of the community and make recommendations to state legislatures on how to improve public policy related to rare diseases.[29]

One of the most pressing issues the RD community currently faces is ensuring that telehealth is appropriately and permanently integrated into our health care system to effectively meet the needs of RD patients and their caregivers. Expanded access to telehealth throughout the pandemic has been particularly beneficial to the estimated 25–30 million Americans living with RDs, reducing their risk of exposure to COVID-19 and helping them better manage their complex health conditions from the safety of home.

Before the COVID-19 pandemic started, we surveyed our RD community and asked about their access to providers. We found that 39% of RD patients travel more than 60 miles for their medical appointments. Therefore, it is no surprise that RD patients have had such a positive reception to expanded telehealth access because of the pandemic. One of NORD's COVID-19 surveys found that, of the 88% of patients who were offered a telehealth appointment, 92% of those who accepted the appointment said it was a positive experience, and 70% would like the option for telehealth for future medical appointments.[30]

Rare disease patients tend to require regular contact with their health care team because their diseases tend to be complex, requiring ongoing monitoring, testing, evaluation, and treatment management.[9] With RD specialists commonly operating out of academic medical centers and children's hospitals—the majority of which are in urban areas—geographically disparate people experience significant barriers to care including transportation and childcare costs and taking time off work or school.[31] We have heard in countless FDA patient-focused drug development meetings of stories where parents of a RD child had to uproot their entire lives, moving cross-country, staying in hotels, and changing jobs to access care.[32] Therefore, access to specialist care is too often limited to those of higher SES who have medical literacy, physician advocates, occupational flexibility, and other privileges.

For the past 2 years, in response to the clear call from RD patients and families in our community surveys, NORD and advocates in our Rare Action Network have worked together and in partnership with other patient groups to advocate to ensure telehealth is available to meet the needs of RD patients and their caregivers.

National Organization for Rare Disorders believes that effectively integrating telehealth into our health care system will lead to better outcomes for RD patients by reducing barriers care, shortening the time it takes to get an accurate diagnosis, and increasing access to providers with RD expertise. In support of these goals, we developed principles[33] to help guide our telehealth policy efforts. To date, many states and private insurance companies have already integrated aspects of telehealth into their plans and programs, yet there is more work to be done.

Both the federal government and states have the power to regulate aspects of telehealth, including telehealth licensure requirements, reimbursement rates, and eligible services. In January of 2022, NORD joined hundreds of organizations in advocating for a pathway to comprehensive telehealth access, considering the eventual end of the federal COVID-19 public health emergency declaration—a circumstance that enabled telehealth expansion at the outset of the pandemic.[34]

National Organization for Rare Disorders also joined a letter to congressional leaders with over 400 organizations supporting permanent telehealth Medicare reforms, as well as collaborating with the ALS Association, the Alliance for Connected Care, and 235 other patient organizations to call on governors to protect patient access to telehealth. From 2020 to 2022, we have advocated through 30 letters, statements, and testimonies for telehealth access and expansion.[2]

There are still barriers to care involving telehealth: low-income, elderly, and rural Americans can lack access to broadband services—whether because the technology infrastructure does not exist in their community, they cannot afford it, or they lack technology literacy to use it in their homes.[35] Therefore, NORD has advocated for the coverage and reimbursement of audio-only telehealth services because it can help bridge this digital divide. National Organization for Rare Disorders mobilized our advocates and the larger RD community to successfully advocate for the inclusion of several telehealth provisions in the omnibus appropriations package passed signed into law in March 2022. These provisions will prevent patients, particularly Medicare beneficiaries, from experiencing a sudden drop in telehealth coverage while we continue to advocate for more comprehensive legislation at the state and federal level. The bill would collect data on how telehealth is improving access to necessary care, even when it is across state lines, which is particularly important for our policy

efforts around ensuring patient access to health care providers practicing in a different state.

CONCLUSION

As an umbrella for hundreds of RD nonprofits, NORD is well situated to observe and learn DEI best practices from these communities, share organizing strategies, and develop new resources synthesizing these learnings to distribute across the entire spectrum of RD organizations. Our overarching goal continues to be to help patient advocacy organizations understand concepts relating to various social determinants of health as well as ways they can help address inequities through diversifying board leadership; reevaluating the design and marketing of their Websites, campaigns, and activities; and encouraging more diverse research participation.

Long-term, this type of programming helps to build strong and culturally competent leaders across the RD community that can continue to mentor and support others—ultimately leading to greater representation for all individuals living with or caring for someone with a RD.

National Organization for Rare Disorders will continue its commitment to understanding barriers to health care in the RD community and lead efforts to spur change by conducting scientific and policy research and studies as well as convening practitioners, leaders from NIH, FDA, patient advocacy organizations, patients, and caregivers in discussions and collaborations to improve access to care for all communities within the RD advocacy space.

References

1. PHA history: 30 milestones in 30 years. Pulmonary Hypertension Association. Accessed March 15, 2022. https://phassociation.org/history/
2. Olson D, Paton P, Simpson J, Dukart B. The PHA story: from kitchen table to internationally respected organization or the history of its foundation, development and growth. National Organization for Rare Disorders. Accessed March 15, 2022. https://2f126cpzgfmjndc42glkg7mg-wpengine.netdna-ssl.com/wp-content/uploads/2017/02/The-PHA-Story-A-Short-History.pdf
3. History of leadership. National Organization for Rare Disorders. Accessed March 15, 2022. https://rarediseases.org/about/what-we-do/history-leadership/
4. Asquith A, Sava L, Harris AB, Radix AE, Pardee DJ, Reisner SL. Patient-centered practices for engaging transgender and gender diverse patients in clinical research studies. *BMC Med Res Methodol*. 2021;21:202. doi:10.1186/s12874-021-01328-4
5. Braveman PA, Kumanyika S, Fielding J, et al. Health disparities and health equity: the issue is justice. *Am J Public Health*. 2011;101(Suppl 1):S149–S155. doi:10.2105/AJPH.2010.300062
6. Valdez R, Ouyang L, Bolen J. Public health and rare diseases: oxymoron no more [published correction appears in *Prev Chronic Dis*. 2016;13:E20]. *Prev Chronic Dis*. 2016;13:E05. doi:10.5888/pcd13.150491
7. Ending the diagnostic odyssey for children with a rare disease: Global Commission year one report. Global Commission to End the Diagnostic Odyssey for Children with a Rare Disease. Published 2019. Accessed March 15, 2022. https://www.globalrarediseasecommission.com/Report
8. Hartin SN, Means JC, Alaimo JT, Younger ST. Expediting rare disease diagnosis: a call to bridge the gap between clinical and functional genomics. *Mol Med*. 2020;26(1):117. doi:10.1186/s10020-020-00244-5
9. Barriers to rare disease diagnosis, care and treatment in the US: a 30-year comparative analysis. National Organization for Rare Disorders, Inc. Published November 19, 2020. Accessed March 29, 2022. https://rarediseases.org/wp-content/uploads/2020/11/NRD-2088-Barriers-30-Yr-Survey-Report_FNL-2.pdf
10. de Souza ÍP, Androlage JS, Bellato R, Barsaglini RA. Doenças genéticas raras com abordagem qualitativa: revisão integrativa da literatura nacional e internacional [A qualitative approach to rare genetic diseases: an integrative review of the national and international literature]. *Cien Saude Colet*. 2019;24(10):3683–3700. doi:10.1590/1413-812320182410.17822019
11. Sirugo G, Williams SM, Tishkoff SA. The missing diversity in human genetic studies. *Cell*. 2019;177(4):1080. doi:10.1016/j.cell.2019.04.032
12. Zvereff VV, Faruki H, Edwards M, Friedman KJ. Cystic fibrosis carrier screening in a North American population. *Genet Med*. 2014;16(7):539–546. doi:10.1038/gim.2013.188
13. Farooq F, Mogayzel PJ, Lanzkron S, Haywood C, Strouse JJ. Comparison of US federal and foundation funding of research for sickle cell disease and cystic fibrosis and factors associated with research productivity. *JAMA Netw Open*. 2020;3(3):e201737. doi:10.1001/jamanetworkopen.2020.1737
14. Healthy people 2030. Office of Disease Prevention and Health Promotion. Accessed March 15, 2022. https://health.gov/healthypeople/objectives-and-data/social-determinants-health
15. Ogundipe F, Mehari A, Gillum R. Disparities in sarcoidosis mortality by region, urbanization, and race in the United States: a multiple cause of death analysis. *Am J Med*. 2019;132(9):1062–1068.e3. doi:10.1016/j.amjmed.2019.03.048
16. Graham GN. Why your ZIP code matters more than your genetic code: promoting healthy outcomes from mother to child. *Breastfeed Med*. 2016;11:396–397. doi:10.1089/bfm.2016.0113
17. Harper LJ, Love G, Singh R, Smith A, Culver DA, Thornton JD. Barriers to care among patients with sarcoidosis: a qualitative study. *Ann Am Thorac Soc*. 2021;18(11):1832–1838. doi:10.1513/AnnalsATS.202011-1467OC
18. Morton DH, Morton CS, Strauss KA, et al. Pediatric medicine and the genetic disorders of the Amish and Mennonite people of Pennsylvania. *Am J Med Genet C Semin Med Genet*. 2003;121C(1):5–17. doi:10.1002/ajmg.c.20002
19. Amish Community Testimony. National Organization for Rare Disorders. Accessed March 15, 2022. https://rarediseases.org/amish-community-testimony/
20. Akers A, Al-Shahi Salman R, Awad IA, et al. Synopsis of guidelines for the clinical management of cerebral cavernous malformations: consensus recommendations based on systematic literature review by the Angioma Alliance Scientific Advisory Board Clinical Experts Panel. *Neurosurgery*. 2017;80(5):665–680. doi:10.1093/neuros/nyx091
21. Heckman BD, Britton AJ. Headache in African Americans: an overlooked disparity. *J Natl Med Assoc*. 2015;107(2):39–45. doi:10.1016/S0027-9684(15)30023-7
22. Manole AK, Forrester VJ, Zlotoff BJ, Hart BL, Morrison LA. Cutaneous findings of familial cerebral cavernous malformation syndrome due to the common Hispanic mutation. *Am J Med Genet A*. 2020;182(5):1066–1072. doi:10.1002/ajmg.a.61519
23. Staff. Foundation for Sarcoidosis Research. Accessed March 15, 2022. https://www.stopsarcoidosis.org/about/staff/
24. Member snapshot. National Organization for Rare Disorders. Accessed March 15, 2022. https://rarediseases.org/for-patient-organizations/membership-profiles/members/
25. Leading with intent: reviewing the state of diversity, equity, and inclusion on nonprofit boards. BoardSource. Published June 2021. Accessed March 15, 2022. https://boardsource.org/wp-content/uploads/2021/06/2021-Leading-with-Intent-DEI-Report.pdf?hsCtaTracking=532d5c05-43be-4e2b-91d1-2ae87fb81017%7C4051e648-43cf-4267-bf08-1a4c8f4b5916
26. Top languages other than English spoken in 1980 and changes in relative rank, 1990–2010.

United States Census Bureau. Updated October 8, 2021. Accessed March 15, 2022. https://www.census.gov/programs-surveys/sis/resources/visualizations/spoken-languages.html
27. NORD Rare Disease Centers of Excellence—find a center. National Organization for Rare Disorders. Accessed March 15, 2022. https://rarediseases.org/centersofexcellence/find-rare-disease-center-of-excellence/
28. Policy issues. National Organization for Rare Disorders. Accessed March 15, 2022. https://rarediseases.org/advocate/policy-priorities/policy-issues/
29. Project RDAC. National Organization for Rare Disorders. Accessed March 15, 2022. https://rarediseases.org/projectrdac/
30. Ensuring access to telehealth for rare diseases. National Organization for Rare Disorders. Published October 15, 2020. Accessed March 15, 2022. https://rarediseases.org/wp-content/uploads/2020/10/NRD-2098-RareInsights-Telehealth-Report-1.pdf
31. Narayanan R. Committing to care, access, equity and research: NORD announces 31 rare disease centers of excellence. National Organization for Rare Disorders. Published November 4, 2020. Accessed April 26, 2022. https://rarediseases.org/committing-to-care-access-equity-and-research-nord-announces-31-rare-disease-centers-of-excellence/
32. External resources and information related to patients' experience. US Food and Drug Administration. Accessed March 15, 2022. https://www.fda.gov/drugs/development-approval-process-drugs/external-resources-and-information-related-patients-experience#collapseOne
33. NORD telehealth principles. National Organization for Rare Disorders. Accessed March 15, 2022. https://rarediseases.org/advocate/policy-priorities/nord-telehealth-principles/
34. NORD telehealth pathway to reform letter to congress. National Organization for Rare Disorders. Accessed March 15, 2022. https://rarediseases.org/wp-content/uploads/2022/01/Telehealth-Pathway-to-Reform-Letter-to-Congress-w-signers-FIN.pdf
35. MacKinney AC, Coburn AF, Lundblad JP, McBride TD, Mueller KJ, Watson SD. Access to rural health care—a literature review and new synthesis. Rural Policy Research Institute. https://rupri.org/wp-content/uploads/20140810-Access-to-Rural-Health-Care-A-Literature-Review-and-New-Synthesis.-RUPRI-Health-Panel.-August-2014-1.pdf