RDDC
RARE DISEASE DIVERSITY COALITION

NORD®
National Organization for Rare Disorders

Inequities in the Rare Disease Community

The Voices of Diverse Patients and Caregivers

June 2024

The Rare Disease Diversity Survey is a collaboration between the Rare Disease Diversity Coalition (RDDC) and the National Organization for Rare Disorders (NORD®).

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About the Survey Sponsors

About Black Women's Health Imperative (BWHI) / Rare Disease Diversity Coalition (RDDC)

The Black Women's Health Imperative (BWHI) is a national non-profit organization dedicated to advancing health equity and social justice for Black women, across the lifespan, through policy, advocacy, education, research, and leadership development. The organization identifies the most pressing health issues that affect the nation's 22 million Black women and girls and invests in the best of the best strategies and organizations that accomplish its goals. You can find more information about BWHI at bwhi.org.

The Rare Disease Diversity Coalition (RDDC) is an initiative launched by BWHI to address the extraordinary challenges faced by historically underrepresented populations with rare disease. RDDC brings together rare disease experts, health and diversity advocates, and industry leaders to identify and advocate for evidence-based solutions to reduce racial disparities in the rare disease community. You can find more information about RDDC at rarediseasediversity.org.

The Rare Disease Diversity Coalition's Patient & Caregiver Working Group (now known as the Patient, Provider and Caregiver Journey Working Group) is comprised of 52 individual rare disease advocates and leaders from rare disease nonprofit organizations, professional societies, medical centers, drug manufacturers, and other key stakeholders.

About the National Organization for Rare Disorders (NORD®)

The National Organization for Rare Disorders (NORD®) is the leading independent advocacy organization representing over 30 million Americans affected by a rare disease. NORD is committed to the identification, treatment, and cure of more than 10,000 rare diseases, of which approximately 95% are still without an FDA-approved treatment or therapy. NORD began as a small group of patient advocates that formed a coalition to unify and mobilize support to pass the Orphan Drug Act of 1983. For more than 40 years, NORD has led the way in voicing the needs of the rare disease community, driving supportive policies and education, and advancing medical research and providing patient and family services for those who need them most. NORD is also home to more than 360 disease-specific member organizations and their communities and collaborates with many other organizations on specific causes of importance to the rare disease community.

Since its establishment, NORD has sought to address issues of health equity for the rare disease community in the United States. Representing patients and families from across 10,000 unique rare diseases has required NORD to focus on outreach and engagement to underserved and often isolated communities to build a unified community that strives to drive public policy, accelerate research and improve patient care.

Learn more about the National Organization for Rare Disorders (NORD®) at rarediseases.org.

Background

There are **more than 10,000 different rare diseases** (1),

of which **95% lack any FDA-approved treatment** (2).

Rare diseases impact small patient populations of **fewer than 200,000 people** in the United States—sometimes far fewer. These smaller patient populations make it **difficult to diagnose and access clinical care and treatments**.

Individuals living with rare diseases commonly face a long and protracted 'diagnostic odyssey.' According to a 2020 NORD study, "Barriers to Rare Disease Diagnosis, Care and Treatment in the US: A 30-Year Comparative Analysis," more than a quarter of rare disease patients spend seven years or more until they receive a correct diagnosis, up from 15% 30 years ago. In addition, more than a third of rare disease patients receive at least one misdiagnosis during their diagnostic journey. Limited medical specialization is likely a driving factor, together with other factors. These factors include complex disease manifestations which affect multiple organ systems, leading to the need for multiple medical specialists. They also include wait times to see a specialist and the need for additional diagnostic testing to rule out or confirm a diagnosis (3).

Across all rare disease communities, delays in diagnosis are a burden and diminish health outcomes. Data demonstrates that diagnostic delays disproportionately affect patients from historically underserved communities, exacerbating long-standing health inequities. At the same time, expeditious access to diagnostic testing is of great importance for individuals with rare diseases as they often face progressive and degenerative symptoms. For instance, according to the same 2020 NORD study, receiving newborn screening significantly increases the odds of rare disease patients being diagnosed in a timely (i.e., within six months) manner (3). Similarly, when individuals affected by rare diseases receive genetic testing, they are more likely to receive a timely diagnosis than patients without, though the difference was not statistically significant in the limited-size data set of the NORD study (3).

Finally, an increasing number of innovative rare disease therapies have narrow treatment windows, so that any delay in diagnosis can – and does – exclude rare disease patients from participating in and potentially benefitting from clinical trials, or from receiving FDA-approved therapies. Given the limited or non-existent alternative treatment options for many rare diseases, such delays can be particularly devastating for patients and families.

Magnifying these challenges to diagnosis, care and treatments, are the added layer of health disparities, which disproportionately impact marginalized communities such as minority ethnic groups, rural populations, LGBTQ+ communities, and low-income Americans, to name a few. The impact of health disparities in the diagnosis, treatment, and clinical outcomes of rare disease patients and the burden on their caregivers have not been systematically investigated across all 10,000 rare diseases and across specific subpopulations.

Executive Summary

In 2022, to tap into the collective expertise and experiences of various stakeholders, the Rare Disease Diversity Coalition (RDDC) convened their Patient and Caregiver Working Group – comprised of 52 rare disease patients, advocates, and leaders from various sectors, including nonprofits, professional societies, medical centers, and drug manufacturers – to identify missing data specific to the patient journey that could provide insights into the unique perspectives and challenges faced by patients and caregivers from historically underrepresented populations. They determined that a national survey would best serve as a pivotal data collection effort to begin identifying and rectifying gaps in rare disease diagnosis, care, and therapy accessibility for diverse and underrepresented patient populations.

In 2022, RDDC and the National Organization for Rare Disorders (NORD) entered into a Memorandum of Agreement to conduct this unprecedented initiative: developing a national survey targeting underrepresented rare disease patients and caregivers across disease areas. The primary objective was to close critical knowledge gaps and gain insights into these individuals' unique perspectives and challenges in accessing and affording health care. By concentrating on underrepresented patients, the intention was to gather essential information that would facilitate a more profound comprehension of existing gaps and barriers in diagnosis, care, and treatment access.

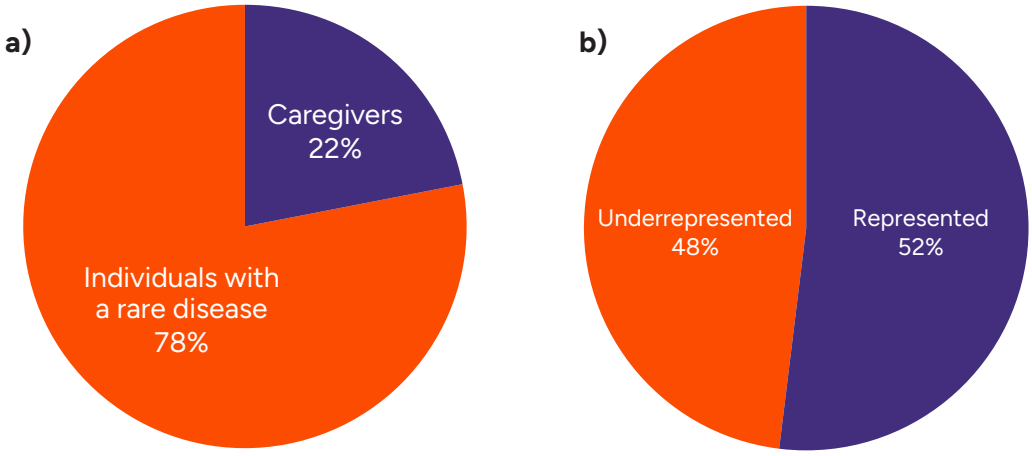
The study aimed to recruit 2,000 respondents, with 60% coming from underrepresented communities based on factors like race, ethnicity, sexual orientation, socioeconomic status, or residence. The recruitment target was surpassed with a final respondent total of 2,848. Among respondents, the majority (78%) were individuals with a rare disease, and the remainder of the surveys were completed by a caregiver for someone with a rare disease (22%). Almost all respondents took the survey in English (99%).

Overall, 1,114 respondents (42%) met the definition of belonging to one or more underrepresented communities; however, 55% of the caregiver respondents were from underrepresented communities. The largest respondent group was Black, Indigenous, or Persons of Color (BIPOC) with 448 respondents. Additionally, 433 respondents were below the federal poverty line, 329 identified as members of the LGBTQ+ community, 270 lived in rural communities based on their reported zip code, and 204 were Hispanic or Latino. Individuals may belong to more than one underrepresented group. For example, among individuals who are BIPOC, 10% are Hispanic, 20% are LGBTQ+, 28% meet the federal definition of poverty, and 9% live in a rural community.

The demographic profile of the respondents can be broken down by gender, education, and ability. Respondents with a rare disease were overwhelmingly female (74%) and caregivers were even more so (88%). Educational attainment varied widely, ranging from some individuals who had not attended school at all to 47% holding a college degree. When asked about ability, 25% of respondents indicated they could not work due to a disability.

FIGURE 1: Distribution of Study Respondents according to a) role in the rare disease community and b) underrepresented or represented community member

Underrepresented groups include BIPOC (Black, Indigenous and People of Color), Hispanic ethnicity, LBGTQ+, Household income below the poverty level or residence in a rural zip code.



Respondents encountered 14 potential factors that may have caused them to delay or forgo care. For seven barriers, over a quarter of the population reported that they led to delays or avoidance of care. These factors included issues related to emotional well-being, financial constraints, limitations stemming from their rare disease, worries about the COVID-19 pandemic, and concerns about interactions with health care providers. The most reported barrier was feeling overwhelmed from managing life, care, or caregiving responsibilities (50% overall). The proportion reporting the barrier was higher among underrepresented communities, with the highest being 72% in the LGBTQ+ community.

When the survey focused on financial barriers to care, 21% of respondents indicated that, during the past year, they could not afford their prescription medications and 20% indicated they could not afford dental care. Among underrepresented respondents, even higher proportions reported an inability to afford care.

INITIAL KEY FINDINGS:

FIGURE 2: Proportion of respondents reporting barriers that led them to delay or forgo care

Have you delayed care or decided not to receive care for any of the following reasons in the past 12 months?

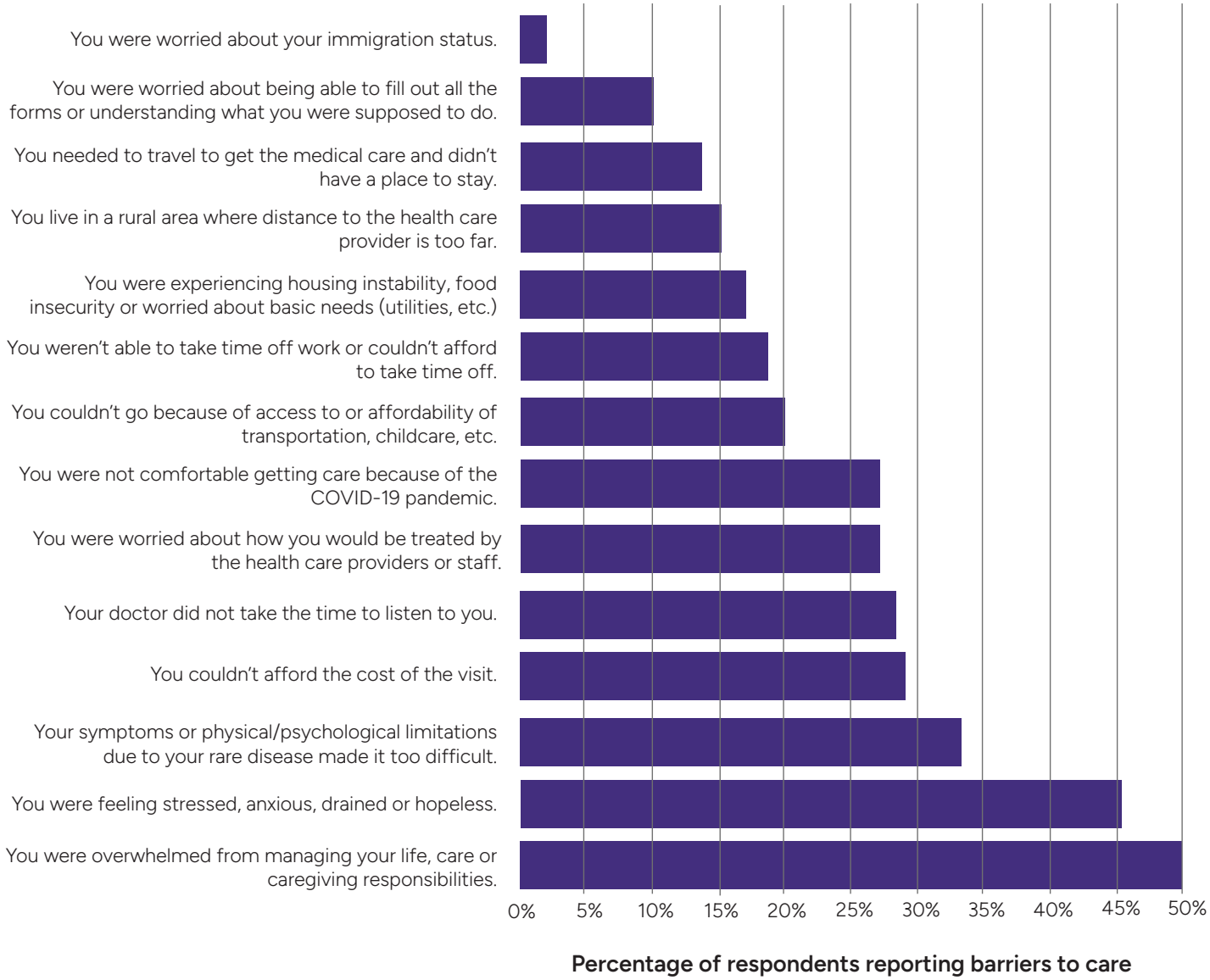
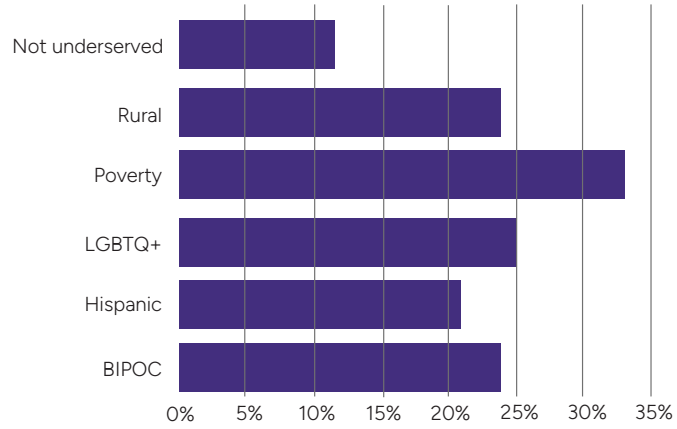
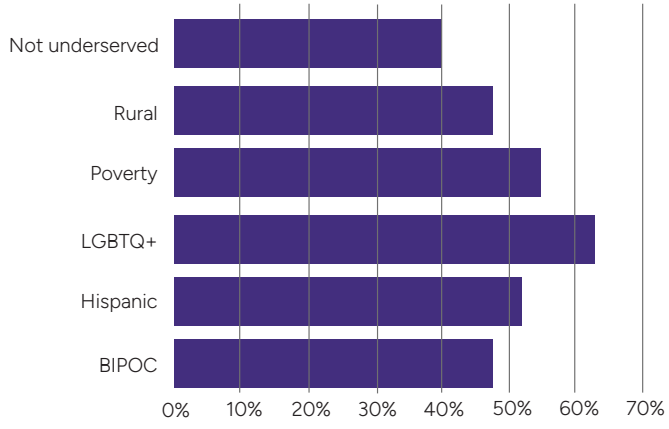


FIGURE 3: Proportion of Respondents indicating they delayed or forgoes care by underrepresented community for the following barriers a) worry about basic needs b) were feeling stressed, anxious, drained or hopeless, c) were overwhelmed from managing life with or caring for someone with a rare disease, d) physical limitations from the rare disease and e) worry about the cost of the visit.

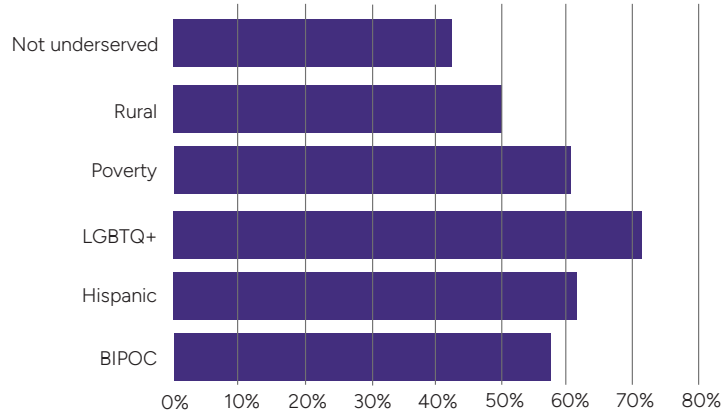
You were experiencing housing instability, food insecurity or worried about basic needs (utilities, etc.)



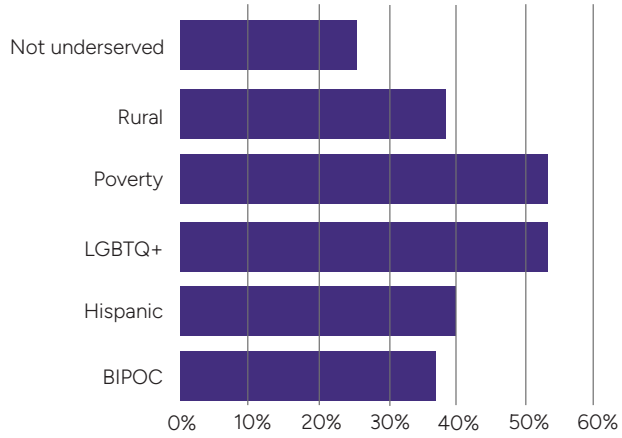
You were feeling stressed, anxious, drained or hopeless



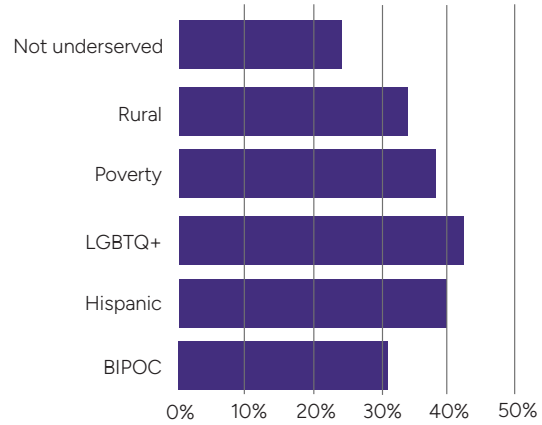
You were overwhelmed from managing your life, care or caregiving responsibilities



Your symptoms or physical/psychological limitations due to your rare disease made it too difficult



You couldn't afford the cost of the visit



Almost all respondents indicated they have had health insurance for the past 12 months (93%). Private insurance provided by employers or obtained through the exchange covered 49% of respondents. At the same time, the remaining individuals relied on public insurance, with Medicare and Medicaid being the most commonly reported options (35% and 16%, respectively). Individuals in underrepresented communities were about three times more likely to report Medicaid as their primary insurance than individuals in represented communities. Medicare denials were reported in 11% of the respondents, and 23% reported Medicaid denials. The proportion of individuals reporting denials was higher in underrepresented communities.

When caregivers were questioned about the effects of their role on their physical, emotional, social, and financial well-being, a significant portion of respondents acknowledged experiencing an impact. Specifically, 85% of respondents agreed with the statement that they often felt physically exhausted, and 71% agreed their life satisfaction has suffered because of caregiving. Caregiver respondents from underserved communities were likelier to report negative impacts on their finances and health than those from historically well-supported and represented communities.

Survey Rationale and Objectives

SURVEY RATIONALE

RDDC and NORD aim to eliminate health disparities for rare disease patients in the United States through evidence-based tools and interventions. However, there is a gap in knowledge and understanding regarding many aspects of the experience of individuals from diverse backgrounds living with rare diseases. Therefore, a national data collection effort focusing on underrepresented patients in the US was critical to provide the information needed to understand gaps and barriers to diagnosis, care, and treatment access.

The overarching goal of the Rare Disease Diversity Survey was to collect information from a diverse sample of individuals with a rare disease and those who care for them to better understand their experiences around accessing diagnostic tools, clinical care, and therapies, and to investigate whether differences are observed across populations.

The rationale underlying the survey was the need to bring the rare disease community together to actively contribute to solutions that remove the barriers to care and treatment for rare disease patients in historically marginalized populations, and support equitable access to education, resources, and advocacy efforts in the rare disease space.

IMMEDIATE AND DISTAL OBJECTIVES

The survey's immediate objective was to conduct a needs assessment and better understand the perceived and actual barriers and gaps for marginalized rare disease patients in accessing diagnosis, clinical care, and treatments. The needs assessment will inform the development of recommendations to address barriers to diagnosis and care among marginalized rare disease patients.

A distal objective is to generate additional questions by RDDC, NORD, and external researchers for subsequent phases of data analysis to assess more complex experiences of sub-groups and to understand differences between the intersectionality of sub-groups. For example, are there differences in gaps and barriers to care of Latinos living in urban cities versus rural areas? Are there differences between LGBTQ+ African Americans versus cisgender, heterosexual African Americans? To what extent does poverty impact Asian Americans and Pacific Islanders in barriers to care? NORD, the RDDC, and other rare disease stakeholders plan on utilizing this data for further analysis for years to come to inform engagement priorities, research areas, program development, and policy recommendations.

REPORT OBJECTIVES

The objectives of this report are to:

- Provide an overview of the approach taken to design and collect data
- Provide a summary analysis of the data collected in the survey
- Inform needs assessment and focus programming to address barriers
- Consider barriers with substantial prevalence (example criterion: reported in above 25% of total respondents)
- Consider barriers that are substantially more prevalent in underrepresented communities (example criterion: absolute difference of 10% or more between represented and underrepresented communities)

Methodology

DESIGN OF SURVEY

With funding and support from RDDC and its Patient & Caregiver Working Group members, NORD coordinated the development, design, implementation, and management of the online survey, *The Diversity in Rare Diseases Survey*. We developed and refined the survey questions over several months and a collaborative, deliberative process. The survey's formulation and review process for national distribution involved meticulous crafting, drawing on insights and recommendations from working group members. This esteemed group collaborated over many months to develop a mixed-method study encompassing qualitative and quantitative questions.

The survey delved into key areas, including demographic information, rare disease attributes, and individual perspectives and experiences across five thematic areas:

- **Diagnostic Journey:** Understanding the challenges and nuances of the journey individuals with rare diseases undertake to receive a diagnosis
- **Health Insurance Status:** Investigating the respondent's current health insurance status, recognizing insurance's pivotal role in accessing healthcare services
- **Healthcare Coverage, Affordability, and Utilization:** Assessing the adequacy of healthcare coverage, affordability of services, and the extent to which individuals utilize available healthcare resources
- **Healthcare Experiences:** Exploring individuals' overall experiences within the healthcare system, identifying strengths and areas for improvement
- **Emotional Health Issues:** Recognizing the emotional toll that rare diseases can take on patients and caregivers, focusing on mental health and well-being

DEFINING UNDERREPRESENTED COMMUNITIES

For this study, we defined underrepresented patients and caregivers by the following criteria:

- Below the federal poverty limit versus above the federal poverty limit
- Black, Indigenous, People of Color (BIPOC) versus non-BIPOC
- Hispanic/Latino versus non-Hispanic/Latino
- LGBTQ+ versus cisgender/heterosexual
- Rural versus urban/suburban

INCLUSION CRITERIA

Inclusivity was a crucial aspect of the study's design, ensuring anyone personally impacted by rare diseases was welcome to participate. This encompassed individuals living with a rare disease, individuals currently undiagnosed but suspected to have a rare disease, and current and former caregivers of individuals with or suspected to have a rare disease.

To foster greater inclusion, the survey instrument was made available entirely in English and Spanish, ensuring individuals from diverse linguistic backgrounds could participate comfortably. All survey messaging, whether in English or Spanish, maintained a sensitive and culturally appropriate tone to resonate with the target audience.

Respondents were required to have access to a smartphone device, desktop, or laptop computer to participate. Most Americans own a smartphone (85%) and a desktop or laptop computer (77%) (4). Even so, we considered options for telephonic survey data collection. We declined to add a telephone option as the increased duration of time required to complete the survey, which already took 20-25 minutes to complete, with added translations, was considered a burden on participants.

SURVEY TECHNOLOGY SELECTION

Several survey software instruments were identified, reviewed, and assessed with considerations for accessibility for the visually impaired, multilingual use, mobile-readiness, and accessibility for those without high-speed internet connections (i.e., fast load times.) Ultimately, SurveyMonkey's platform met these requirements and was selected to host the survey questions. The platform also meets WCAG 2.1[1] accessibility standards and 508 compliance.[2]

ANONYMITY OF RESPONDENTS

Data was collected anonymously to foster open and honest responses and remove certain barriers to participation, including concerns about the privacy of personal health information and disclosure.

There was consideration of possible financial incentives to encourage survey participation and decrease survey dropout rates, but prior experience demonstrated that monetary incentives raise the risk of fraudulent respondents, which would ultimately undermine data quality. Incentivizing results would also require that individuals leave their name and contact information, excluding individuals who fear online data collection, privacy breaches, and immigration concerns.

BETA-TESTING THE PLATFORM

The survey questions were beta-tested in English and Spanish by 13 patients, caregivers, and rare disease advocates to test for health literacy levels, language sensitivity, cultural biases, accessibility for physical disabilities, and limited-broadband users. The beta testers ensured the survey platform was laptop and mobile accessible and that the survey questions were manageable, culturally sensitive, and comprehensible. Beta testers also tested the branching logic of questions and the overall flow of the survey. They measured the time it took to complete the survey and allowed us to gauge survey fatigue and the potential for survey dropouts. Their feedback was integrated into the survey design and several questions were edited as a result.

[1] WCAG2 is a set of technical guidelines for making web content more accessible. It includes a set of criteria to test for success.

[2] Section 508 is a United States federal law that requires all electronic and information technology used by the federal government to be accessible to people with disabilities.

Recruitment Methods

Our multifaceted and inclusive approach to data collection underscored RDDC and NORD's commitment to capturing diverse perspectives. By utilizing a mix of online platforms, community events, and targeted outreach efforts, the survey sought to amplify the voices of those often underrepresented in health care research. The thoughtful combination of anonymity assurance, language accessibility, and strategic distribution channels contributed to the success of this pioneering national survey. Details on the recruitment methods used are described below.

Recruitment Timeline

The survey launched on February 14, 2023, and closed after four months on June 15, 2023.

Media Relations

Prior to the survey launch, a joint press release was issued and distributed on the national PR Newswire to announce the collaboration and survey objectives (5). The press release was intended to elicit interest in the survey and engage high-level leaders and other stakeholders to prepare for the survey launch.

Email and Website Distribution

To maximize engagement and reach, the survey was frequently promoted by NORD through targeted emails to patients and caregivers, patient advocates, volunteers, patient organizations, patient advocacy group leaders, community-based organizations, biopharmaceutical representatives, health care providers, patients and caregivers. In addition to direct promotion, we requested the support of partners to distribute the survey to their networks.

In total, NORD emails with the survey were sent to 33,621 people, with 15,583 opened. This 46% open rate and 9.5% click rate exceed industry averages. Additionally, RDDC emailed the survey to 404 subscribers in its biweekly newsletter over the course of eight weeks. These emails were opened 44 times.

A popup window advertisement was also created and used on the NORD homepage throughout the duration of the recruitment window to direct visitors to the survey.

Social Media Outreach

A social media toolkit was created for partner promotion across Facebook, Twitter, Instagram, and LinkedIn. The toolkit was distributed to 85 members of RDDC's coalition and 52 leaders in their Patient and Caregiver working group. In addition, NORD distributed the social media toolkit to 560 leaders of nonprofit rare disease patient advocacy organizations in its membership, 1,200 community-based organizations, and 40 academic medical centers through its NORD Rare Disease Centers of Excellence network. The toolkits were also hosted on NORD's website, available in English and Spanish (6, 7).

From February through June, the survey received promotional posts on NORD's LinkedIn, Instagram, Facebook, and X/Twitter. Table 1 collects NORD's engagement metrics for these social media posts by platform. Overall, LinkedIn had the highest click-through rate (CTR), which is our best measure of audience engagement, followed by Facebook and Instagram. Twitter had the lowest level of engagement. This could point to insights about which platforms are best for researchers to reach individuals living with rare diseases of diverse backgrounds. Limitations of this analysis include that LinkedIn posts got significantly more NORD staff engagement than other platforms and posts were in English only, though some included Spanish graphics. Sample social media advertisements can be viewed in the Appendix.

Table 1: Social Media Metrics for Survey Promotion on NORD-owned Digital Channels

Platform	Post Views	Survey Clicks	Click-through-rate (CTR)	Platform Follower Count
LinkedIn	8,673	159	1.8%	27,770
Instagram	7,605	114	1.5%	11,600
Facebook	6,555	102	1.6%	80,000
Twitter	4,476	27	0.6%	40,600
Total	**27,309**	**402**	**1.5%**	**160,000**

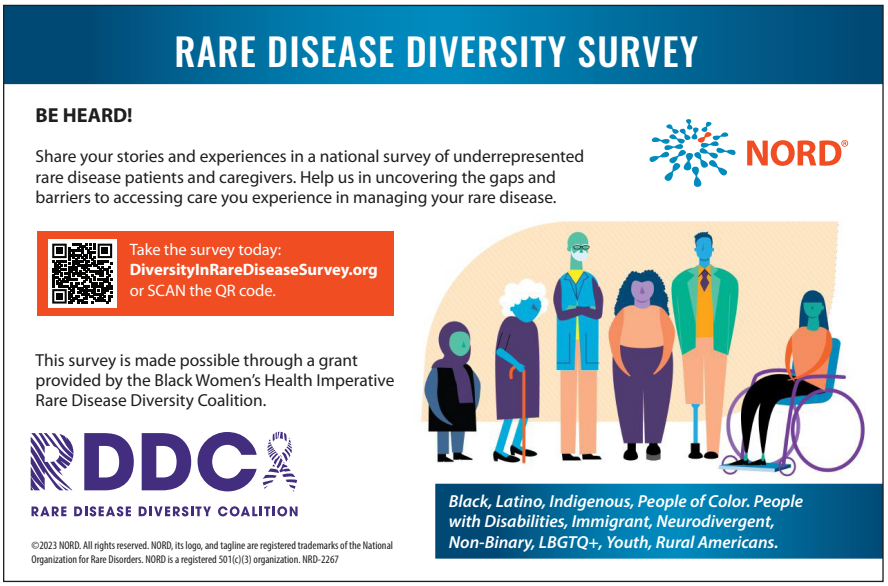
Partnering with Ethnic-Based Advocacy Groups and Rare Disease Leaders

Collaboration with advocacy groups focused on specific ethnicities and disorders, as well as major hospitals, provided an avenue to connect with underrepresented communities who would not be reached through traditional channels.

Successful outreach was conducted to Black Health Matters, the Asian American Pacific Islander Health Forum, Gillette Children's Hospital, Sick Cells, Looms for Lupus, National Organization for African Americans with Cystic Fibrosis, the Scleroderma Research Foundation, the George Washington University School of Medicine, Children's National Hospital, and others.

Community Engagement

Promotional flyers in English and Spanish were disseminated at professional medical society and health conferences, health fairs, stakeholder meetings, legislative events, Rare Disease Day events, and at other in-person promotional opportunities. We announced calls to request bundles of 50-200 postcards for promotion, with nearly 2,000 being distributed. Through this effort, potential participants were reached in person in 14 states: Arkansas, Arizona, Connecticut, DC, Florida, Maryland, Michigan, North Carolina, New Jersey, Ohio, Oklahoma, Tennessee, Texas, and Wisconsin.



The flyers were made available in several formats, in English and Spanish, and were offered for use online, by email, and in person. The promotional flyers were offered in two sizes: one 8½ x 11-inch flyer for distribution via email and in doctor's offices and one 4 x 8 inch postcard, which were printed and distributed to rare disease organizations, coalitions, advocates, drug companies, medical clinics and media partners for distribution at in-person events. They included a QR code linking to the survey.

Paid Advertising

The Black Women's Health Network graciously donated high-impact digital billboard advertising space in Times Square, New York City, advertising the survey in English and Spanish. Times Square is ostensibly one of the most ubiquitous advertising locations in the country and averages nearly 380,000 pedestrians and another 115,000 drivers and passengers passing through each day; Times Square signage advertising can expect nearly 1.5 million impressions each day (8). RDDC received complimentary placement on a Times Square digital billboard in New York City during the month of May. This billboard was wrapped around the building corner of an intersection to face two different sides of the street.

Additionally, during the last two weeks of the survey distribution (June 1-15, 2023), paid advertisements were run across Google, YouTube, and Google's display network. We curated a list of rare diseases that disproportionately impact communities of color[3] and rolled out additional advertisements targeting users searching or reading articles about these specific rare disease terms, as well as more general terms.

> ***Google Search and YouTube ads were viewed 405,436 times and clicked-through 2,587 times, resulting in more than 2,500 visits to the Survey form at a relatively low cost of $1.13 per click.***

Given the high bar of taking a 20-minute survey and the text space constraints for the ads to guide users from the articles they were reading to the survey, we believe the reach and engagement of the paid advertisements indicates a very strong result.

Survey Results

RECRUITMENT RESULTS

The study aimed to recruit 2,000 respondents, 60% from underrepresented communities based on race, ethnicity, sexual orientation, socioeconomic status, or rural residence. The recruitment target was surpassed with a final total of 2,848 respondents (Table 2). Among respondents, the majority (78%) were individuals with a rare disease, and the remainder of the surveys were completed by a caregiver for someone with a rare disease (22%) (Table 2). More than half of the participants completed the survey in February, the first month it was available, with steady increases in the total number of respondents observed between March and June.

It is important to note that this was an anonymous survey. This simplified the study enrollment process and likely made participants more comfortable with reporting sensitive information, but as a result, participants could not be validated and their eligibility as a member of the rare disease community could not be verified.

DEMOGRAPHICS OF SURVEY RESPONDENTS

The study participants were primarily adults and almost 90% of individuals with a rare disease were aged 18 or older. Individuals with a rare disease and caregivers were overwhelmingly female (74% and 88%, respectively) (Table 3). Most of the caregiver respondents are currently caregiving (84%) (Table 2). Among individuals with a rare disease, 8% were African American or Black, 4% were from an Indigenous population, 5% were Asian or Middle Eastern, and the remaining 83% were White. There was a range in educational attainment from those who had not attended school to 47% of the individuals with a rare disease having a college degree. Half of the individuals with a rare disease were married or living with a partner. Slightly less than half, 44%, were employed or students, whereas about a quarter of respondents indicated that they were unable to work due to disability. Almost all respondents took the survey in English (99%) (Table 3).

[3] The list was not all-inclusive and the resources used to curate the diseases varied from peer-reviewed publications to NORD's online organizational database and Rare Disease Reports.

Overall, 1,259 respondents (48%) met the definition of belonging to one or more underrepresented communities. Of note, 65% of the caregiver respondents were from underrepresented communities. The largest respondent group was individuals who are Black, Indigenous or Persons of Color (BIPOC) with 448 respondents. Additionally, there were 433 respondents who were below the federal poverty line, 329 individuals who identified as members of the LGBTQ+ community, 270 living in rural communities based on their reported zip code, and 204 Hispanic/Latino respondents. Individuals may belong to more than one underrepresented group. For example, among respondents who are BIPOC, 19% are Hispanic/Latino, 14% are LGBTQ+, 19% meet the federal definition of poverty, and 3% live in a rural community (Table 4).

Survey respondents are significantly impacted by their rare diseases. The majority (59%) have one rare disease, while 26% report having two or more (Table 5). Overall, the rare diseases represented by respondents demonstrate impacts across all major organ systems and, for many individuals, impacts across multiple organ systems. More than 40% of respondents indicated their disease impacted their brain, immune system, or muscular system. Furthermore, rare diseases influence every aspect of daily life: 60% of individuals have mobility issues, 45% have difficulty performing complex tasks, 44% report their disease affects their ability to work or attend school, and 65% find their ability to enjoy activities with friends and family is hindered. Individuals from underrepresented communities reported greater impacts on all organ systems and all life activities.(Table 6).

RESULTS OF BARRIERS ASSESSMENT

Respondents were presented 14 different potential factors that may have caused them to delay or forgo care. These factors were related to emotional health, financial barriers, limitations resulting from their rare disease, concerns about the COVID-19 pandemic, and concerns about interactions with providers. For seven of the barriers, more than 25% of the population indicated that it caused them to delay or forgo care. The most reported barrier was feeling overwhelmed from managing life, care or caregiving responsibilities (50% overall, 72% in the LGBTQ+ community). For all barriers, the proportion reporting the barrier was higher among underrepresented communities. There was variability across underrepresented communities of the proportion of individuals who reported a barrier. For nine of the barriers, the highest proportion of individuals reporting was among the LGBTQ+ community. These barriers provided some validation of the definitions of underserved community as 43% of those in a rural community noted distance from providers as a barrier (as compared to 15% of the total population) and 31% of individuals below the federal poverty limit noted housing instability and food insecurity as barriers (compared to 17% overall) (Table 9).

In the section on financial barriers to care, 21% of respondents indicated that during the past year they were unable to afford their prescription medications and 20% indicated they could not afford dental care. Among underrepresented respondents, there were even more substantial proportions reporting an inability to afford care, including 25% unable to afford prescription medications, 24% unable to afford dental care, 21% unable to afford eyeglasses and hearing aids, 19% unable to see a specialist and 18% unable to afford mental health care (Table 10).

DIAGNOSTIC EXPERIENCE

The vast majority of respondents indicated they have been diagnosed with a rare disease, with only 5% indicating they were currently undiagnosed. The majority of respondents, 83%, were diagnosed because they experienced symptoms. Overall, 45% of the respondents had received genetic testing and 33% had met with a genetic counselor. Of those who reported not receiving testing, 85% indicated the reason was because it was not offered. Interestingly, rates of genetic testing, visits with genetic counselors, and applications to the Undiagnosed Disease Network were slightly higher among respondents from underrepresented populations; however, rates remain too low overall, indicating a need for increased access across all demographics (Table 7).

HEALTH INSURANCE AND ACCESS TO CARE

Almost all of the respondents indicated they have had health insurance for the past 12 months (93%). Private insurance through employers or the exchange was the coverage for 49% of respondents and the remainder had public insurance, with Medicare and Medicaid being the most reported (35% and 16% respectively). Individuals in underrepresented communities were about three times more likely to report Medicaid as their primary insurance than individuals in well-represented communities. While about a quarter of respondents did not have costs for insurance premiums, 42% of the respondents spend $250 or more each month on health insurance. In addition, individuals incurred additional health care costs with 22% of respondents indicating costs over $1,000 dollars per month. Medicare denials were reported by 11% of

the respondents and Medicaid denials were reported by 23% of the respondents. The proportion of individuals reporting denials was higher in the underrepresented communities (Table 8).

CAREGIVER ASSESSMENTS

When caregivers were asked about their experiences as a caregiver and the impacts on them physically, emotionally, socially and financially, a substantial percentage of the respondents indicated an impact. Specifically, 85% of respondents agreed with the sentence that they often felt physically exhausted and 71% agreed that their life satisfaction has suffered because of caregiving. Respondents from underrepresented communities were more likely to report negative impacts on their finances (69% versus 56%) and health (72% versus 58%) than respondents from well-represented communities (Table 11).

Conclusion

In conclusion, this collaborative effort in developing and implementing a comprehensive national survey marks a significant stride toward addressing health disparities in rare diseases. The insights gained from this survey can inform evidence-based strategies, interventions, and policies to eliminate barriers to diagnosis, care, and treatment access for underrepresented patient populations. As the data analysis continues to unfold, we are poised to play a pivotal role in shaping a more equitable health care landscape for those with rare diseases, fostering a future where every individual, regardless of background, can access the care they need and deserve.

The Rare Disease Diversity Survey highlights the power of bringing multiple stakeholders together to co-design a survey and collectively use their connections and channels to publicize it and support recruitment. Involvement of the broad range of stakeholders in RDDC's Patient and Caregiver Working Group ensured the survey addressed a well-rounded set of key questions that were meaningful to the community. Through the collective efforts of all stakeholders, the survey was able to surpass its goal of 2,000 respondents, demonstrating the effectiveness of a united approach in reaching a large audience. The extensive dissemination strategies included social media campaigns, email newsletters, community events, and partnerships with various organizations, all contributing to the high response rate.

However, even with widespread dissemination, it remained a challenge to recruit participants from underserved communities. Despite targeted outreach efforts, such as collaborating with local community leaders, translating materials into multiple languages, and providing incentives, these communities were still underrepresented in the survey responses. This underscores the persistent barriers to participation that underserved populations face, such as limited access to information, distrust in research, and socioeconomic constraints.

The findings from the Rare Disease Diversity Survey underscore the necessity of ongoing efforts to engage and include diverse populations in research. Addressing these challenges requires continuous, targeted outreach and the development of trust and rapport within these communities. Moving forward, the insights gained from this survey can inform future strategies to enhance inclusivity and ensure that the voices of all individuals affected by rare diseases are heard and considered in research and policy-making.

Recommendations

The Rare Disease Diversity Survey provides a much-needed foundation of data to understand the current experiences of individuals living with a rare disease, and particularly those from underserved communities. Based on this initial examination of the data, there are several areas of need where action is appropriate. In many cases, the data indicates that the entire rare disease community is struggling, and effective action can be broad. However, there are also signals that burdens and barriers are more substantial among those in underrepresented communities, and in these cases interventions and actions need to be more targeted. These recommendations should serve as examples, with the understanding that additional recommendations will emerge from subsequent analysis and discussions.

INCREASE ACCESS TO GENETIC COUNSELING

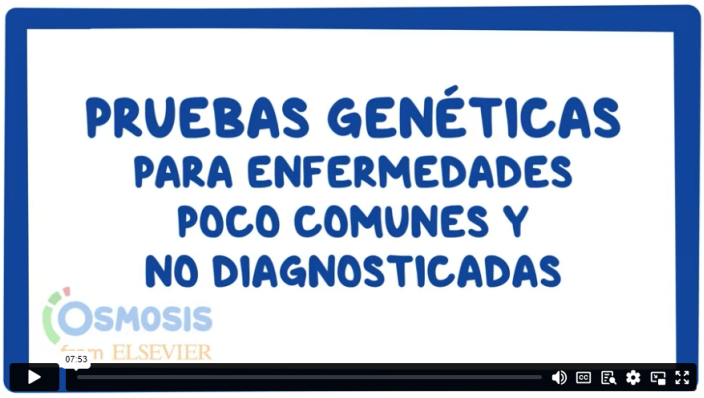
Research suggests that approximately 72% of rare diseases are genetic in origin (9), however, fewer than half of our respondents have received genetic testing. Of those who did not receive testing, 85% were never offered it. There is an urgent need to ensure all individuals have access to affordable options for genetic counseling as part of their diagnostic process, particularly in communities historically underserved by the health care system. Clinicians across the health care system must then be properly educated about these options and when and how to offer them.

EXISTING RESOURCES:

- NORD has explainer videos on "**Genetic Testing for Rare and Undiagnosed Diseases**" in English and Spanish.

Genetic Testing for Rare and Undiagnosed Diseases - Video

Genetic Testing for Rare and Undiagnosed Diseases (Spanish) - Video



EXPAND INSURANCE GUIDANCE AND COVERAGE

While most respondents have health insurance, a substantial proportion indicated their insurance did not allow them to access the providers, medication, procedures, and telehealth services needed to manage their health. Access issues were reported more frequently among respondents from underrepresented communities. This highlights an opportunity for financial navigators to support rare disease patients, particularly in regions where more underrepresented people live, to educate them about existing options for co-pay assistance. We must advocate for more financial assistance programs where needs are not being met and for state governments to uphold the commitments of the Affordable Care Act to protect patients from inadequate health plans. Lastly, we can and must reform state and federal policies that create unnecessary bureaucratic barriers to care, particularly as it relates to care across state lines.

EXISTING RESOURCES:

- NORD Patient Assistance Programs offer financial assistance for patients with certain diagnoses.
- NORD's State Resource Center includes a directory of organizations and programs offering insurance support state by state.
- The Patient Advocate Foundation (PAF) operates a Co-Pay Relief Program to assist with out-of-pocket medical costs. (Accessible in Spanish here.)
- The Patient Advocate Foundation (PAF) also operates a Rare Disease CareLine to help rare patients navigate financial and logistical barriers.

UNDERSTAND AND CORRECT FOR THE PROFOUND IMPACTS OF POVERTY ON HEALTH ACCESS

The finding that more than 20% of people in underrepresented communities delayed or skipped care because they lacked basic needs of food and shelter is deeply concerning and warrants attention and action. Until the root causes of poverty and income inequality are addressed—issues which should be understood as critical to the rare disease community—there is a dire need for proactive outreach and financial and administrative assistance for low-income individuals and families with rare diseases. Assistance with food and shelter should be considered as components of care for these members of the rare disease community and be provided by organizations with means in the space. There is an opportunity for industry partners to reexamine their existing support and consider new support that addresses these social determinants of health as a means of improving access to medical care.

EXISTING RESOURCES:

- RDDC has a guide to Social Security Disability Benefits for Rare Disease Patients that explains which disorders qualify for disability benefits and compassionate allowances and how to apply for them.
- NORD's Patient Assistance Programs offer financial assistance for some non-medical costs of living, as well as emergency relief, depending on an individual's diagnosis.
- NORD's State Resource Center includes a directory of state programs offering assistance with housing and other costs of living, medical equipment and adaptive technology, nutrition and wellness, and more.
- FindHelp.org connects patients to free or reduced-cost resources like food, housing, financial assistance, and health care near them.
- The U.S. Department of Health and Human Services Administration for Children & Families has an Office of Community Services (OCS) that operates a Low Income Home Energy Assistance Program (LIHEAP) to assist families with energy bills and weatherization repairs.

ADDRESS THE MENTAL HEALTH CRISIS FACING PATIENTS AND CAREGIVERS

It is common for rare disease patients to delay or forgo care because they feel stressed, anxious, drained, or hopeless or are overwhelmed from managing life, care or caregiving responsibilities. More than half of respondents indicated each of these as barriers to care, with a higher proportion among those in underrepresented communities. There is a need to develop and disseminate culturally informed mental health and stress management education, tools, and programs for the rare disease community with the ultimate goal of increasing access to therapy and respite services for rare patients and caregivers. For rare caregivers, specifically, there is a significant need for advocacy, programs, and tools to help them effectively manage their own health and finances. Some such programs exist, such as NORD's Caregiver Respite program, but new programs must be implemented to meet the observed demand. Overall, there is an opportunity for advocacy organizations to seek funding from industry partners and foundations to provide psychosocial support from social workers for the rare disease patients and families they serve.

EXISTING RESOURCES:

- NORD's Caregiver Respite Program offers assistance for unpaid rare disease caregivers to attend events or simply take time off.
- NORD's State Resource Center includes a directory of programs offering mental health assistance state by state.
- Raregivers, formerly Angel Aid, offers community groups and supportive tools for rare disease caregivers globally.

PROMOTE ENGAGEMENT AND INCLUSION IN DAILY LIFE

Individuals' daily lives are substantially impacted by their rare diseases, with 60% of respondents reporting impacts on mobility and 65% reporting impacts on abilities to enjoy activities with family and friends. There is an opportunity and an imperative to build bridges that lessen the isolation and loneliness of living with a rare disease. We recommend educating the wider, non-rare community about the realities of living with a rare disease and strategies to be more inclusive at school, in the workplace, and at social events. Where possible, develop new programs and resources to facilitate social interaction and community-building. Rare disease community events such as NORD's Living Rare, Living Stronger Patient and Family Forum offer financial, travel, and lodging assistance at the community level to overcome these barriers to engagement in daily life, and can be used as a model for other organizations considering scholarships to their meetings.

EXISTING RESOURCES:

- RDDC created a Diversity, Equity and Inclusion (DEI) Organizational Readiness Quiz for patient organizations to assess how effectively they are meeting the needs of diverse populations, extending beyond race and ethnicity to include age, gender, geographic location, sexual orientation, disability, and more.
- The annual Living Rare, Living Stronger® NORD Patient and Family Forum is free for rare disease patients and families and offers scholarships for first-time attendees. Conference topics often cover how to advocate for yourself in the healthcare system, how to coordinate your medical care team, and how to get involved in rare disease research and advocacy, as well as general networking with others in the community.
- Our Odyssey is an inclusive platform connecting young adults living with rare diseases and chronic conditions with virtual and live events.

ALIGN THESE RECOMMENDATIONS WITH EXISTING AND FUTURE POLICY CAMPAIGNS

The survey data underscores the urgent need for evidence-based policies and programs to mitigate inequities facing underrepresented communities with rare diseases. Collaborative efforts with community organizations, health care providers, and governmental bodies (such as state Rare Disease Advisory Councils, or RDACs) can catalyze targeted interventions at national and state levels. Aligning survey data with existing policy and advocacy campaigns, such as the expansion of Medicaid or telehealth services, presents opportunities to strengthen the voice of underrepresented communities in active policy deliberations, ensuring their voices and experiences are not just included but considered. It is recommended that the data be cross-referenced against existing public policy efforts to identify potential advocacy partners and campaigns for which it can be useful, and such work is already underway at RDDC and NORD.

EXISTING RESOURCES:

- RDDC maintains a list of **policy initiatives** pertaining to health equity in rare diseases, as well as **recommendations** for policies to improve diversity in clinical trials. Organizations and individuals seeking to get involved with the coalition can **sign up here**.
- NORD maintains a list of its **public policy positions** and actively mobilizes the community to advocate for policies to improve the lives of rare disease patients at the federal and state level through its **Rare Action Network®**.
- NORD publishes an annual **State Report Card®** assessing each U.S. state's progress on nine issues of importance to the rare community, including Medicaid eligibility, medical nutrition, newborn screening, quality of state-regulated insurance, telehealth access, prescription out-of-pocket drug costs, the establishment of Rare Disease Advisory Councils, and more.

CONTINUE ASSESSMENT AND RECOMMENDATIONS

This analysis sets the stage for future endeavors, including deeper analysis that can inform the development of a comprehensive needs assessment. It is imperative that future activities embark on an exploration of sub-groups within underrepresented communities, delving into greater analysis of the intersections of social determinants of health included in this data. Additionally, there are opportunities to conduct cross-sectional analysis to explore and investigate potential associations across data elements (e.g. the impact of the type of health insurance on reported access barriers, or of educational attainment and language on health literacy as a barrier to care). This thorough assessment will unearth nuanced differences between sub-groups, enabling the formulation of tailored solutions vetted by community members themselves. By identifying new variables and considerations, future surveys can refine knowledge gaps, inform policy development and education initiatives, and support new services.

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Appendix

CLICK HERE TO VIEW ALL SURVEY QUESTIONS

DATA SUMMARY TABLES

Table 2: Overall Responses

	Count
Total	2848
Individuals with a Rare Disease	2208
Caregivers for Individual with a rare disease Current Previous Alive Deceased No answer/skipped	640 540 89 22 49 11

Table 3: Participant and Caregiver Demographics

	Individuals with a Rare Disease	Caregivers
	Count (%)	**Count (%)**
Age		
<2	20 (1%)	0
2 - <6	67 (3%)	0
6 - <12	84 (3%)	0
12 - <18	106 (4)	0
18 - <30	292 (11%)	35 (6%)
30 - <40	287 (11%)	126 (22%)
40 - <50	379 (15%)	158 (28%)
50 - <60	495 (19%)	116 (21%)
60 - <70	508 (20%)	86 (15%)
70 +	354 (14%)	38 (7%)
No answer/skipped	256	81
Sex		
Male	647 (25%)	68 (12%)
Female	1917 (74%)	497 (88%)
Intersex	S	0
None of these describe me	S	0
Prefer not to answer	17	9
No answer/skipped	251	66
Gender		
Male	639 (25%)	62 (11%)
Female	1826 (72%)	493 (88%)
Non-binary	52 (2%)	S
Transgender	S	S
Self-describe	S	0
Prefer not to answer	40	6
No answer/skipped	270	75

Table 3: Participant and Caregiver Demographics *(continued)*

	Individuals with a Rare Disease	Caregivers
	Count (%)	**Count (%)**
Sexuality		
Straight	2054 (88%)	494 (94%)
Gay	42 (2%)	S
Lesbian	52 (2%)	S
Bisexual	134 (6%)	13 (2%)
Self-describe	61 (3%)	S
Prefer not to answer	173	23
No answer/skipped	332	91
Rare		
Black, African American	210 (8%)	33 (6%)
American Indian / Alaska Native / Native Hawaiian, Pacific Islander	98 (4%)	13 (2%)
Asian / Middle Eastern / North African	122 (5%)	32 (6%)
White	2108 (83%)	485 (86%)
Prefer not to answer	66	18
No answer/skipped	244	59
Ethnicity		
Hispanic	182 (7%)	66 (11%)
Non-Hispanic	2422 (93%)	515 (89%)
No answer/skipped	244	59
Educational Attainment		
Never attended school or only attended kindergarten	86 (1%)	S
Grades 1 – 4 (Primary)	73 (3%)	S
Grades 5 – 8 (Middle school)	60 (2%)	S
Grades 9 through 11 (Some high school)	99 (4%)	S
Grade 12 or GED (High school graduate)	278 (11%)	51 (9%)
1 to 3 years after high school (Some college, Associate's degree, or technical school)	690 (28%)	126 (23%)
College 4 years or more (College graduate)	608 (25%)	164 (30%)
Advanced degree (Master's, Doctorate, etc.)	548 (22%)	179 (33%)
Prefer not to answer	27	6
No answer/skipped	368	101
Marital Status		
Married	1063 (44%)	381 (74%)
Divorced	321 (13%)	49 (9%)
Widowed	110 (5%)	17 (3%)
Separated	42 (2%)	12 (2%)
Never married	718 (30%)	35 (7%)
Living with partner	147 (6%)	24 (4%)
Prefer not to answer	62	16
No answer/skipped	385	106

Table 4: Definition of Underrepresented Communities

	Count (%)
BIPOC	448 (18%)
Hispanic Ethnicity	204 (8%)
LGBTQ+	329 (14%)
Household income below poverty level	433 (17%)
Rural Residents	270 (13%)
Total Unique Individuals	1259 (48%)
Examples of Intersectionality BIPOC Only BIPOC + Hispanic BIPOC + LGBTQ+ BIPOC +Below poverty level BIPOC + Rural Residence	 248 (55%) 39 (9%) 63 (14%) 84 (19%) 14 (3%)

Table 5: Diagnosis of Rare Disease(s)

	Count (%)	Underrepresented Respondents	Represented Respondents
Number of Rare Diseases			
1	1562 (59%)	726 (58%)	836 (61%)
2	356 (13%)	176 (14%)	180 (13%)
3	148 (6)	72 (6%)	76 (6%)
4	40 (3)	42 (3%)	28 (2%)
5	95 (4)	56 (5%)	39 (3%)
No answer/skipped	401 (15%)	187 (15%)	214 (16%)
Undiagnosed	105 (5%)	66 (7%)	39 (4%)

Table 6: Characteristics of Rare Diseases

	Total Respondents	Underrepresented Respondents	Represented Respondents
	Count (%)	**Count (%)**	**Count (%)**
Organ Systems Impacted (multi-select)			
Brain	948 (43%)	495 (46%)	453 (40%)
Heart	571 (26%)	303 (28%)	268 (24%)
Blood	460 (21%)	234 (22%)	226 (20%)
Lungs	674 (30%)	338 (31%)	336 (30%)
Gastro-intestinal system	948 (43%)	503 (46%)	445 (39%)
Kidney	474 (21%)	255 (23%)	219 (19%)
Immune System	965 (44%)	487 (45%)	478 (42%)
Bones	574 (26%)	325 (30%)	249 (22%)
Muscles	973 (44%)	513 (47%)	460 (41%)
Skin	695 (31%)	385 (36%)	310 (28%)
Cancer / tumors	308 (14%)	151 (14%)	157 (14%)
Sexual and reproductive systems	283 (13%)	174 (16%)	109 (10%)
Impact of Rare Disease on Life			
Ability to walk and move	1302 (60%)	682 (63%)	620 (57%)
Ability to see	485 (22%)	270 (25%)	215 (20%)
Ability to talk and communicate	569 (26%)	327 (30%)	242 (22%)
Ability to hear	269 (12%)	152 (14%)	117 (11%)
Ability to eat, dress, bathe	700 (32%)	408 (38%)	292 (27%)
Ability to do complex tasks	983 (45%)	547 (51%)	436 (40%)
Ability to remember things	817 (38%)	448 (42%)	369 (34%)
Ability to attend school or work	958 (44%)	537 (50%)	421 (38%)
Ability to enjoy activities with friends and family	1409 (65%)	733 (68%)	676 (62%)
Ability to engage in intimate and sexual relationships	728 (33%)	401 (37%)	327 (30%)

Table 7: Diagnostic Journey

	Total Respondents	Underrepresented Respondents	Represented Respondents
	Count (%)	**Count (%)**	**Count (%)**
Mode of Diagnosis			
Asymptomatic Newborn Screening Routine medical care Family member diagnosed Unknown	375 (17%) 63 (17%) 98 (27%) 79 (21%) 129 (35%)	170 (16%) 33 (20%) 38 (23%) 28 (17%) 66 (40%)	205 (18%) 30 (15%) 60 (29%) 51 (25%) 63 (31%)
Symptomatic	1811 (83%)	887 (84%)	924 (82%)
Genetic Testing Received			
Yes	1019 (45%)	541 (49%)	478 (42%)
No	994 (44%)	444 (40%)	550 (47%)
Genetic testing was not offered	834 (85%)	372 (85%)	462 (85%)
Do not know	232 (10%)	114 (10%)	118 (10%)
Met with a genetic counselor			
Yes	732 (33%)	392 (36%)	340 (30%)
No	1429 (64%)	663 (61%)	766 (67%)
Do not know	71 (3%)	38 (3%)	33 (3%)
Undiagnosed who have not applied to the UDN	85 (89%)	53 (84%)	32 (97%)

Table 8: Health Insurance and Attributes

	Total Respondents	Underrepresented Respondents	Represented Respondents
	Count (%)	**Count (%)**	**Count (%)**
Health Insurance			
Insured past 12 months	2016 (93%)	941 (90%)	1075 (96%)
Insured some of past 12 months	70 (3%)	48 (5%)	22 (2%)
Uninsured past 12 months	82 (4%)	56 (5%)	26 (2%)
Current Primary Health Insurance			
Non-Employment based	184 (9%)	100 (10%)	84 (8%)
Private Insurance Employment-based Private insurance	800 (40%)	373 (39%)	427 (40%)
Medicare	704 (35%)	255 (26%)	449 (42%)
Medicaid	274 (16%)	197 (20%)	77 (7%)
TRICARE	30 (1%) S	19 (2%) S	11 (1%) S
Veterans Affairs (VA)	S	S	S
Indian Health Service	S	S	S
Don't know	23 (1%)	16 (2%)	7 (1%)
Prefer not to answer	30	10	20
Monthly cost for health insurance			
0	465 (27%)	286 (35%)	180 (20%)
1<$100	148 (9%)	72 (9%)	76 (8%)
$100 - <$250	399 (23%)	162 (20%)	237 (26%)
$250 - <$500	386 (22%)	156 (19%)	230 (25%)
$500+	339 (20%)	142 (17%)	197 (21%)
Monthly total health care costs			
$0	134 (7%)	79 (8%)	55 (5%)
$1 - $49	153 (8%)	71 (7%)	82 (8%)
$50 - $99	214 (11%)	85 (9%)	129 (12%)
$100 - $249	433 (22%)	197 (20%)	236 (23%)
$250 - $499	384 (19%)	182 (19%)	202 (20%)
$500 - $999	259 (13%)	142 (15%)	117 (10%)
$1000+	436 (22%)	221 (22%)	215 (21%)
Insurance denials			
Medicare	211 (11%)	126 (13%)	85 (8%)
Medicaid	442 (23%)	249 (26%)	193 (19%)
Insurance Attributes (% reporting always or mostly)			
Find doctors in network	1416 (54%)	604 (48%)	812 (59%)
Get referrals to specialists	1564 (60%)	715 (57%)	849 (62%)
Get approval for procedures, medicines and foods	1360 (52%)	602 (48%)	758 (55%)
Receive telehealth	1114 (44%)	535 (42%)	609 (44%)
Receive reimbursement for supplemental costs	265 (10%)	128 (10%)	137 (10%)

Table 9: Percentage reporting delaying or forgoing care

	Total	BIPOC	Hispanic	LGBTQ+	Poverty	Rural
You were feeling stressed, anxious, drained or hopeless	45%	48%	53%	64%	55%	48%
You were overwhelmed from managing your life, care or caregiving responsibilities	50%	57%	63%	72%	61%	50%
You couldn't afford the cost of the visit	29%	31%	39%	42%	38%	34%
You couldn't go because of access to or affordability of transportation, childcare, etc.	21%	29%	25%	34%	32%	27%
You weren't able to take time off work or couldn't afford to take time off	18%	21%	26%	32%	21%	17%
You were experiencing housing instability, food insecurity or worried about basic needs (utilities, etc.)	17%	24%	21%	25%	31%	23%
You live in a rural area where distance to the health care provider is too far	15%	16%	15%	21%	25%	43%
You needed to travel to get the medical care and didn't have a place to stay	13%	19%	17%	20%	24%	21%
You were worried about your immigration status	1%	3%	1%	1%	2%	0%
Your symptoms or physical/psychological limitations due to your rare disease made it too difficult	33%	37%	40%	54%	54%	38%
You were not comfortable getting care because of the COVID-19 pandemic	27%	33%	31%	44%	35%	25%
You were worried about how you would be treated by the health care providers or staff	27%	34%	31%	51%	32%	24%
You were worried about being able to fill out all the forms or understanding what you were supposed to do	10%	11%	12%	18%	16%	9%
Your doctor did not take the time to listen to you	29%	32%	35%	44%	36%	27%

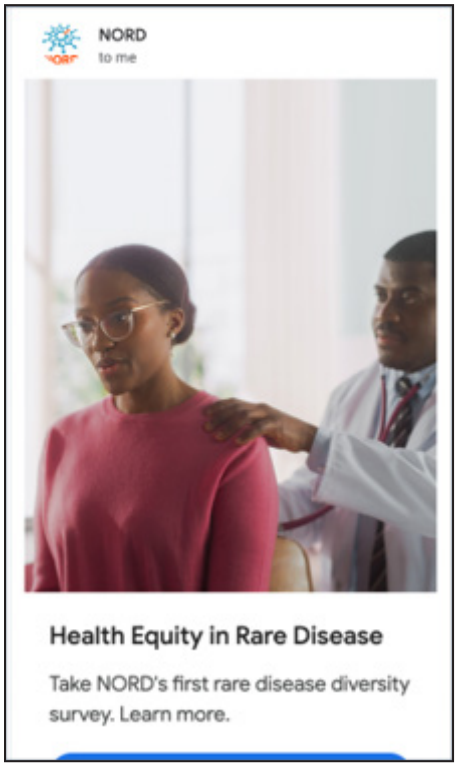
Table 10: Financial barriers to care – Inability to afford within the past 12 months

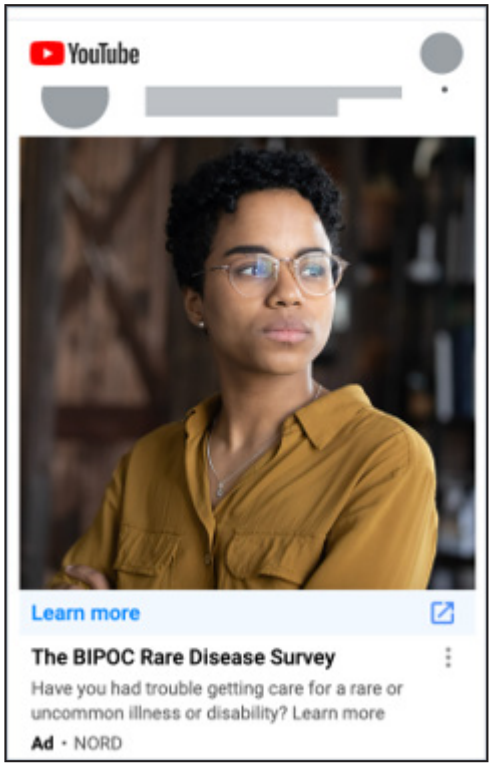
	Total Respondents	Underrepresented Respondents	Represented Respondents
	Count (%)	**Count (%)**	**Count (%)**
Prescription Medicines	549 (21%)	299 (24%)	250 (18%)
Medically necessary food / supplements	306 (12%)	188 (15%)	118 (9%)
Mental health care or counseling	334 (13%)	206 (16%)	128 (9%)
Emergency care	132 (5%)	91 (7%)	41 (3%)
Dental care (including check ups)	511 (20%)	284 (23%)	227 (17%)
Eyeglasses / hearing aids	420 (16%)	245 (19%)	175 (13%)
Mobility aids (wheelchairs etc)	212 (8%)	136 (11%)	76 (6%)
To see a regular doctor or general health provider	157 (6%)	102 (8%)	55 (4%)
To see a specialist	355 (14%)	224 (18%)	131 (10%)
Follow-up care	204 (8%)	128 (10%)	76 (6%)

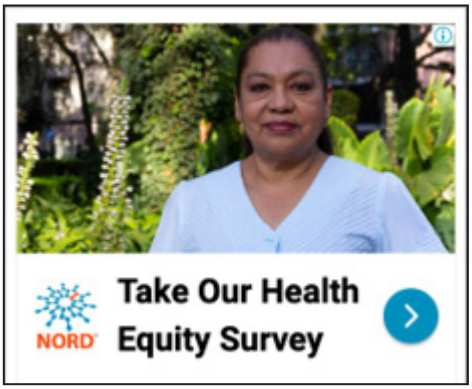
Table 11: Caregiver Assessments of their Experiences

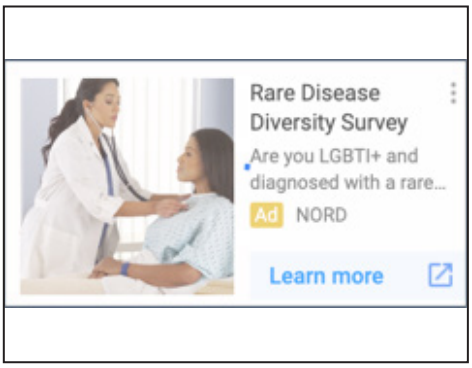
	Percent of Caregivers that Agree or Strongly Agree with these statements		
	Total Respondents	Underrepresented Respondents	Represented Respondents
My life satisfaction has suffered because of the care.	224 (71%)	145 (70%)	79 (72%)
I often feel physically exhausted.	270 (85%)	176 (85%)	94 (85%)
From time to time, I wish I could "run away" from the situation I am in.	198 (62%)	127 (62%)	71 (64%)
Sometimes I don't really feel like "myself" as before.	241 (76%)	154 (75%)	87 (78%)
Since I have been a caregiver my financial situation has decreased.	205 (65%)	142 (69%)	63 (56%)
My health is affected by the care situation.	211 (67%)	146 (72%)	65 (58%)
The care takes a lot of my own strength.	258 (81%)	171 (83%)	87 (78%)
I feel torn between the demands of my environment (such as family) and the demands of the care.	208 (66%)	140 (68%)	68 (61%)
I am worried about my future because of the care I give.	181 (57%)	127 (62%)	54 (48%)
My relationships with other family members, relatives, friends, and acquaintances are suffering as a result of the care.	194 (61%)	127 (62%)	67 (60%)

Appendix B: Sample Advertisements

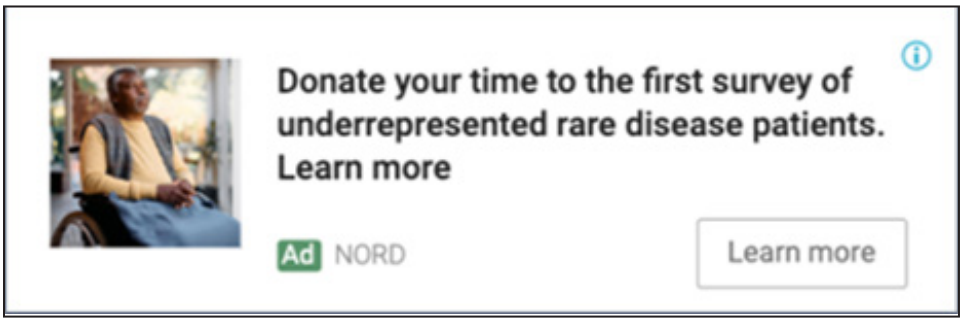








Appendix B: Sample Advertisements (continued)



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