RDDC

RARE DISEASE DIVERSITY COALITION

EXECUTIVE SUMMARY:
Addressing Health Disparities in Rare Diseases through Comprehensive Data Collection

Background:

In 2020, as the devastating COVID-19 pandemic highlighted the additional struggles of patients with rare diseases and people of color, the Black Women's Health Imperative brought together rare disease experts, patient organizations, health and diversity advocates, and industry leaders with a deep knowledge of the medical, industry, regulatory, and cultural challenges facing historically marginalized populations with rare diseases came together and established the Rare Disease Diversity Coalition (RDDC).

There are more than 7,000 different rare diseases, of which 95% lack any FDA-approved treatment.[1] Rare diseases impact small patient populations of fewer than 200,000 in the United States—sometimes far fewer. These smaller patient populations make diagnosing and accessing clinical care and treatments difficult.

Individuals living with rare diseases commonly already face a protracted 'diagnostic odyssey.' According to the National Organization for Rare Disorders (NORD®), more than 1 in 4 rare disease patients spend seven years or more until they receive a correct diagnosis, up from 15% thirty years ago.[2] In addition, more than 1 in 3 rare disease patients receive at least one misdiagnosis during their diagnostic journey.[3]

RDDC believes firmly in collaboration and leveraging the input of stakeholders, including rare disease patients and families, who experience daily the struggles of these issues; patient organizations, who represent and unite those most affected; clinicians and researchers, who drive understanding and evidence across the medical community; private-sector groups, which play a crucial role in supporting research; and policymakers, who drive changes in government policy that lead to progress. To tap into the collective expertise and experiences of various stakeholders, RDDC convened their Patient and Caregiver workgroup in 2022 to identify missing data specific to the patient journey that would gain insights into the unique perspectives and challenges faced by patients and caregivers from historically underrepresented populations, impacted by rare diseases for those concerned by accessing and affording healthcare.

1 https://rarediseases.org/wp-content/uploads/2022/10/NORD-Avalere-Report-2021_FNL-1.pdfhttps://rarediseases.org/wp-content/uploads/2022/10/NORD-Avalere-Report-2021_FNL-1.pdf
2 https://rarediseases.org/wp-content/uploads/2022/10/NRD-2088-Barriers-30-Yr-Survey-Report_FNL-2.pdf.
3 https://rarediseases.org/wp-content/uploads/2022/10/NRD-2088-Barriers-30-Yr-Survey-Report_FNL-2.pdf

Collaborating with the NORD® Patient and Caregiver workgroup embarked on an unprecedented initiative – developing a national survey targeting underrepresented rare disease patients and caregivers. The primary objective is to gain profound insights into these individuals' unique perspectives and challenges in accessing and affording healthcare. As two esteemed organizations committed to addressing rare disease disparities, RDDC and NORD aim to identify and rectify gaps in rare disease diagnosis, care, and therapy accessibility for diverse and underrepresented patient populations.

Purpose:

This groundbreaking endeavor aims to identify evidence-based tools and interventions to eliminate health disparities among rare disease patients, particularly those from underrepresented backgrounds. Recognizing a critical gap in knowledge and understanding of the experiences of individuals from diverse backgrounds living with rare diseases, RDDC and NORD have prioritized focusing on underrepresented patients. By concentrating on underrepresented patients, the intention is to gather essential information that will facilitate a more profound comprehension of existing gaps and barriers in diagnosis, care, and treatment access. The workgroup determined a national survey would best serve as a pivotal data collection effort.

Approach:

The survey's formulation and review process for national distribution involved meticulous crafting, drawing on insights and recommendations from the Rare Disease Diversity Coalition's Patient and Caregiver Working Group. This esteemed working group comprises 52 rare disease patients, advocates, and leaders from various sectors, including nonprofit organizations, professional societies, medical centers, and drug manufacturers. The collaborative effort resulted in 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 several themes:

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

The inclusivity of the survey was a crucial aspect, designed to welcome participation from anyone personally impacted by rare diseases. This encompassed individuals with diagnosed rare diseases, those currently undiagnosed but suspected to have a rare disease, and current and former caregivers of individuals with or suspected to have a rare disease.

Data Collection: Conducting a Groundbreaking National Survey

The success of any data study hinges on the effectiveness of its data collection process. In the case of the first-of-its-kind national survey targeting underrepresented individuals with rare diseases and caregivers, the RDDC and the NORD employed a comprehensive and inclusive data collection strategy. The following outlines the key aspects of this groundbreaking endeavor:

Survey Platform and Duration:

- The survey was administered using the SurveyMonkey platform, offering a user-friendly interface for participants.
- The data collection timeframe extended from February 14 to June 15, 2023, providing a four-month window for individuals to contribute their valuable insights.

Language Accessibility:

- Recognizing the importance of inclusivity, the survey was made available in English and Spanish, ensuring that 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.

Anonymous Data Collection:

- To encourage open and honest responses, the survey embraced a commitment to anonymity. Participants could share their experiences without concerns about the disclosure of personal information.

Distribution Channels:

- **Email Distribution:** The survey reached tens of thousands of potential participants through targeted email distribution within the extensive networks of RDDC and NORD. This included individuals directly impacted by rare diseases and caregivers.
- **Ethnic-Based Advocacy Groups:** Collaboration with advocacy groups focused on specific ethnicities provided an avenue to connect with underrepresented communities. This approach enabled engagement with individuals who typically wouldn't be reached through traditional channels.
- **Community Engagement:** Postcard promotions at exhibits during health conferences and grassroots community health fairs nationwide served as an on-the-ground strategy to capture the attention of potential participants within their local communities.

Social Media Outreach:

- **Multi-Stakeholder Social Media Channels:** Extensive promotion across various social media channels leveraged the reach of multi-stakeholder networks. This approach broadened the survey's visibility, reaching diverse audiences connected to rare disease advocacy, healthcare, and related fields.
- **Paid Google Ads:** Strategic investment in paid Google ads expanded the survey's reach to hundreds of thousands. This targeted approach ensured visibility among individuals searching for rare disease-related information and resources.

This 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 healthcare research. The thoughtful combination of language accessibility, anonymity assurance, and strategic distribution channels contributed to the success of this pioneering national survey.

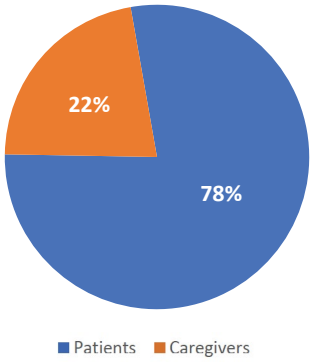
Results:

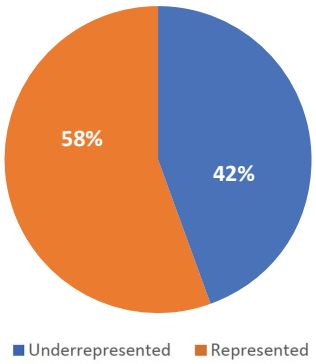
The study aimed to recruit 2,000 respondents, with 60% coming from underrepresented communities based on factors like race, ethnicity, sexual orientation, socioeconomic status, or residence. The recruitment target was surpassed with a final respondent total of 2,848. Among respondents, the majority (78%) 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, 1114 (42%) respondents 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. 433 respondents were below the federal poverty line, 329 individuals identified as members of the LGBTQ+ community, 270 lived in rural communities based on their reported zip code, and 204 were Hispanic respondents. 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 breakdown for gender, education, and ability:

- Individuals with a rare disease and caregivers were overwhelmingly female (74% and 88%, respectively).
- The educational attainment varied widely, ranging from those who had not attended school at all to 47% holding a college degree.
- 25% of respondents indicated they could not work due to a disability.





Total Respondents	2848
Individuals living with a rare disease	2,208 (78%)
Caregivers	640 (22%)

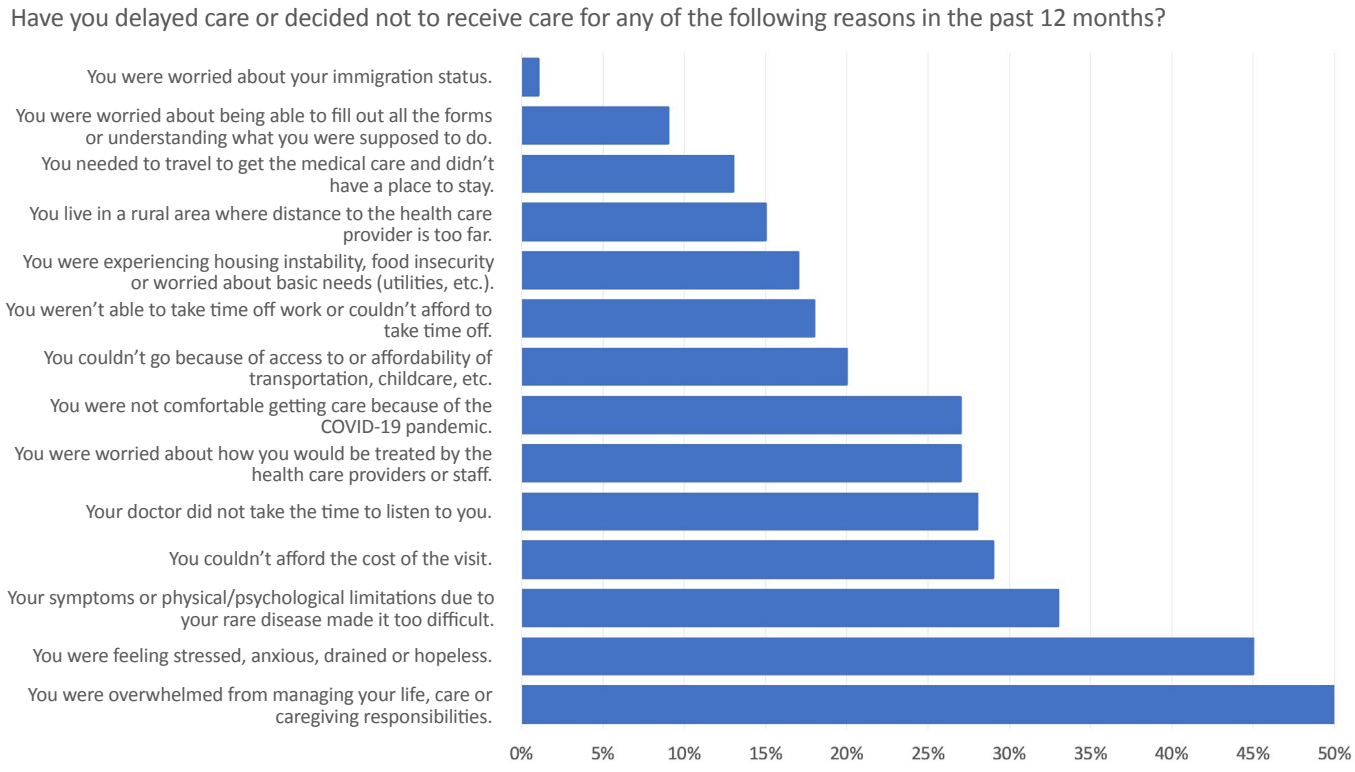
Underrepresented Groups	
BIPOC (Black, Indigenous, POC)	Hispanic ethnicity
Household income below federal poverty limit	Residence in a rural zip code

Respondents:

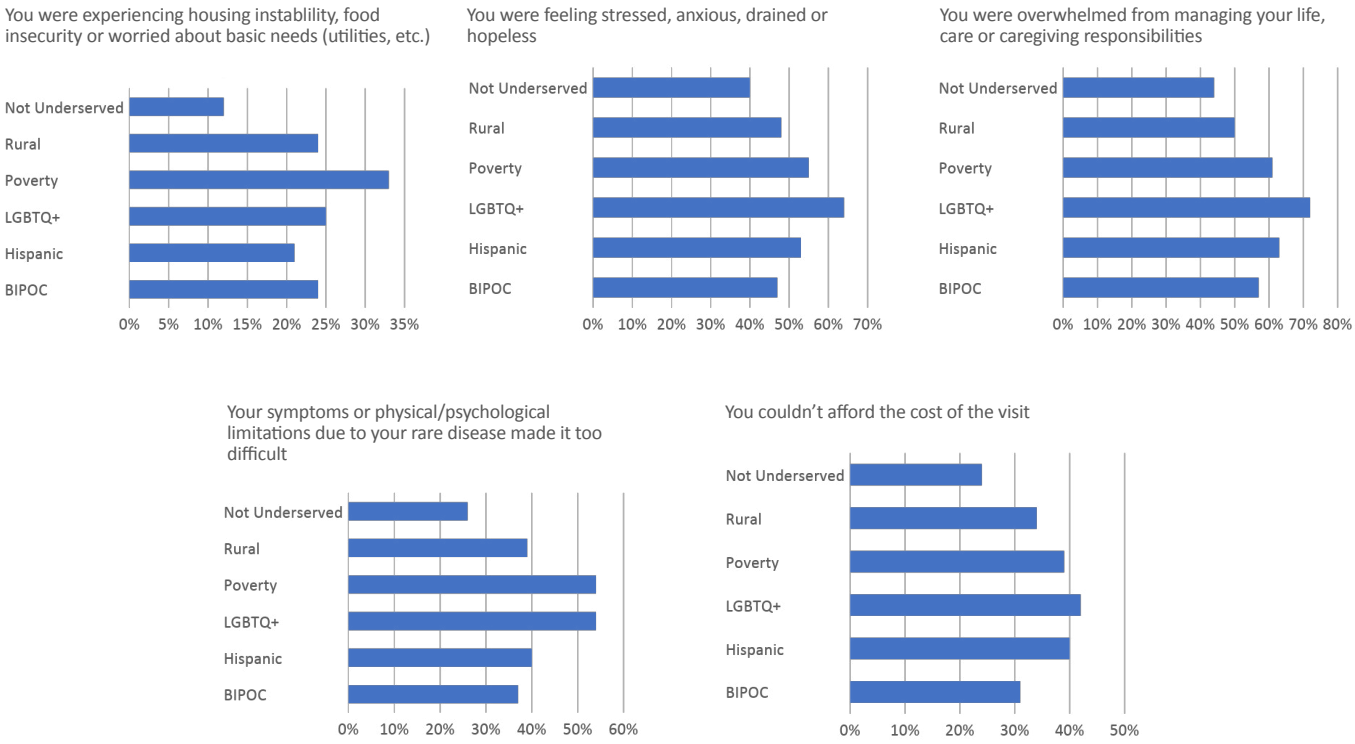
Respondents encountered 14 potential factors that may have caused them to delay or forgo care. For seven barriers, over a quarter of the population reported that they led to delays or avoidance of care. These factors included issues related to emotional well-being, financial constraints, limitations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

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, substantial proportions were reporting an inability to afford care.

Initial Key Findings:



Sample of disparities between populations:



Almost all respondents indicated that 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 the underrepresented communities.

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. 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 that their life satisfaction has suffered because of caregiving.

Conclusion

In conclusion, our 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 unfolds, we are poised to play a pivotal role in shaping a more equitable healthcare landscape for those with rare diseases, fostering a future where every individual, regardless of background, can access the care they need and deserve.

A comprehensive summary of the report will be released in mid-2024.
Initial findings suggest the following:

- Traditional responses to access to care offered by some partners may have shifted and do not always support diverse, vulnerable populations living with rare disease.
- Several of the challenges/gaps addressed in diverse rare communities also apply to the non-rare communities.
- Preliminary evidence supports previously made assumptions regarding gaps and challenges within this community, substantiating these claims.
- Caregivers from underserved communities are more likely to experience negative financial and health impacts than those from historically well-supported communities.