Rare Disease:

Addressing Inequities in Care and Improving Quality

February 13, 2025

Webinar Logistics

- **Send us your questions.** Use the "Q&A" feature at the bottom of the screen to send speakers your questions throughout today's presentation.
- **Today's forum is being recorded.** A copy of the slides and recording will be shared with PQA members within a week.
- **Give us your feedback.** A quick survey will launch at the end of today's webinar.

Presenters

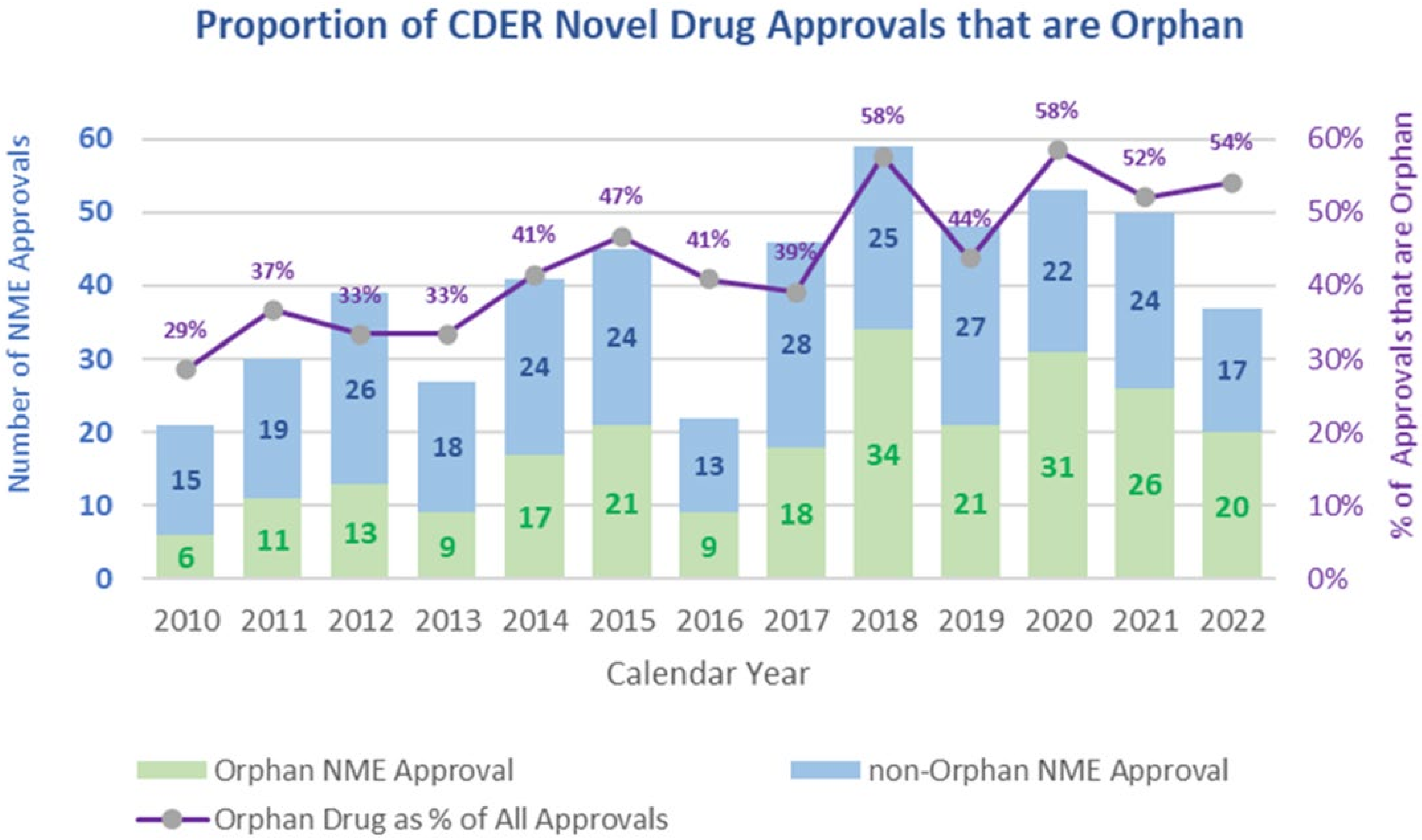
Jocelyn Cooper
Program Manager, Rare Disease Diversity Coalition
Black Women's Health Imperative

Richard Schmitz
Chief Engagement Officer
PQA

Rare Disease and Medication Use

- A disease or condition that affects fewer than **200,000** people in the United States
- There are more than **7,000** rare diseases
- Together they affect more than **30 million** Americans
- Economic burden of rare disease reached nearly **$1 trillion** in 2019
- Only about **5%** of rare diseases have treatments that are approved by the FDA

Increase in Rare Disease Medications



PQA Convenes Goals

PQAConvenes
Quality Medication Use
in Rare Disease

- Highlight the importance of quality in rare disease medication use
- Discuss the barriers to medication access
- Focus on the care that surrounds a rare disease medication prescription
- Build a foundation for consensus on quality issues

PQA Convenes Perspectives

- Patients, Caregivers and Patient Advocates
- Specialty Pharmacies and Providers
- Biopharmaceutical Industry Organizations
- Public and Private Payers

Key Quality Insights from the Panels

- Care coordination is critical for organizing, managing and supporting a patient’s complex needs across multiple care providers and specialties.
- Access to patient health records enables specialty pharmacies to help patients navigate medication approval processes, coordinate care and provide needed services.
- Ongoing collection of rich data that captures the full patient experience is critical to establishing the quality and value of rare disease medications.
- Care management is the backbone of quality.

Barriers to Accessing Care

PQA Convenes Participants

1. Was overwhelmed from managing life, care or caregiving responsibilities
2. Couldn’t afford the cost of the visit
3. Live in a rural area where distance to the health care provider is too far
4. Wasn’t able to take time off work or couldn’t afford to take time off
5. Couldn’t go because of access to or affordability of transportation, childcare, etc.

Patients’ & Caregivers’

1. Was overwhelmed from managing life, care or caregiving responsibilities
2. Felt stressed, anxious, drained or hopeless
3. Symptoms or physical/psychological limitations due to rare disease made it too difficult
4. Couldn’t afford the cost of the visit
5. Doctors did not take the time to listen

Download the Report

PQA

PQA CONVENES: QUALITY MEDICATION USE IN RARE DISEASE

A Report on Stakeholder Perspectives on What Constitutes Quality

Key Quality Insight

Care coordination is critical for organizing, managing and supporting a patient's complex needs across multiple care providers and specialties.

Patients with rare disease see multiple providers, and the burden is on them to track who they've seen and when, which tests have been conducted, which medications they currently take, and which medications or treatments they have tried in the past. Patients must be their own advocates – ensuring providers have all the relevant history, asking questions, probing for more information and searching for providers who see them as whole people and help them maintain as much normalcy as possible. "Whole person" care is a patient-centered approach that accounts for an individual's physical, behavioral, psychological and social needs.

High-quality experience and positive outcomes

Having affordable, high-quality health insurance is the most beneficial tool in achieving positive outcomes and a positive patient experience. It's also paramount to find the right doctor. Patients need doctors who believe them without dismissing symptoms. They need providers who are willing to look for the right diagnosis and willing to work to find which treatments are most effective.

It's also important for patients and their caregivers to receive education on their condition so they can be better advocates. Patients who have the best experience are often a "squeaky wheel" who doesn't take no for an answer. Creating ambassador progra and awareness campaigns can provide a sense of community to patients and a way to connect with others who have the same condition.

Patients, especially the underinsured, would benefit f improved and expanded access to genetic tests, wh could speed up diagnosis and help avoid unnecessary or unsuccessful treatments. There also are significant monetary costs to patients, when they are not accura diagnosed, as ineffective medications can lower or del improvements to quality of life and daily functioning.

Even when patients have health care training or knowledge, it can be difficult to navigate care and benefits. This underscores the challenges of our health care system and patients' need for partners and a supportive system to achieve the best outcomes.

Health equity

Health literacy and limited time and resources are major barriers to high-quality care for many rare disea patients. Simply being able to take time off from work travel for appointments is not feasible for many patient Patient advocates and knowledgeable case workers are needed to support these individuals.

For populations that speak little to no English, who need translators or who may be undocumented, there are additional barriers to care. Building trust with these communities is the first step in breaking down barriers. Patients need to feel safe and believe you are working in their best interests.

Patients need to be met where they are, by people who look at the journey from the patient's perspective rather than the clinical perspective. The "what" and "why" need to be explained. If the care plan doesn't make sense to the patient, the individual is much less likely to participate in follow-up care. Additionally, patients who have lower levels of education often have their concerns dismissed by physicians and are less equipped to "fight" for a different answer.

PUBLIC AND PRIVATE PAYER PERSPECTIVES

The fourth and final panel featured payer perspectives with insights from individuals with experience leading commercial, Medicare, Medicaid and state programs and plans and managing prescription drug coverage for health plans. Payers are focused on medication outcomes and the impact of therapies on total costs of care.

Key indicators of quality and value

When medications are prescribed, payers want to make sure the medications are clinically appropriate, that patients are provided supportive monitoring and care management, and have access to additional support to ensure their needs are routinely being met.

When looking for quality and value, payers are looking for safety and efficacy. Payers want to know:

- did the medication do what it was prescribed to do?
- what are those outcomes?
- how does it impact the total cost of care?

Ideally the medications are showing a long-term reduction to the cost of health care.

Payers are always looking at new drugs coming to the market, the clinical implications and impact on whole-person care, as well as for holes in the evidence. Often, there is limited data for new medications or medications for small populations, which makes evaluations of quality challenging.

Public and Private Payer Perspectives
panelists

DAVID KELLEY | MD, MPA, Chief Medical Officer, Pennsylvania Department of Human Services Office of Medical Assistance Programs (Medicaid)

DRAKE REITER | PharmD, Director, Pharmacy Operations, Priority Health

JAY TRAN | PharmD, MHA, Vice President, Clinical Operations, Capital Rx

MICAH COST | PharmD, MS, CAE, Chief Executive Officer, PQA (moderator)

Specialized services, support services and care management

Specialized services and support play a key role in helping patients have a high-quality experience and the best possible outcomes. Payers use a variety of strategies and partners to support complex case management for individuals with rare disease.

Payers look for community-based programs, including waiver programs, special needs units for children and wrap around services to better assist their members to ensure their needs are met. This care management approach includes a focus on maximizing in-network care opportunities, while also tracking issues such as the distance between patients and their pharmacies and providers, which can impact use and outcomes.

Payer size often impacts the approach to case management. Large payers may have the staff and internal structure to provide direct care management, while smaller payers may rely on external services to provide care management or leverage provider partnerships to assist.

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www.pqaalliance.org/assets/docs/PQA_Convenes_Rare_Disease_Report.pdf

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RDDC
THE RARE DISEASE DIVERSITY COALITION

Inequities in the Rare Disease Community

BLACK WOMEN'S
HEALTH IMPERATIVE

Special Thanks to Today's Host

PQA

QUALITY MEDICATION USE

BWHI/RDDC At a Glance

BLACK WOMEN'S
HEALTH IMPERATIVE

RDDC™ Vision Statement

To live in a world where rare disease care is the standard of care for all, ensuring that patients receive equitable access to diagnosis, treatment, and support.

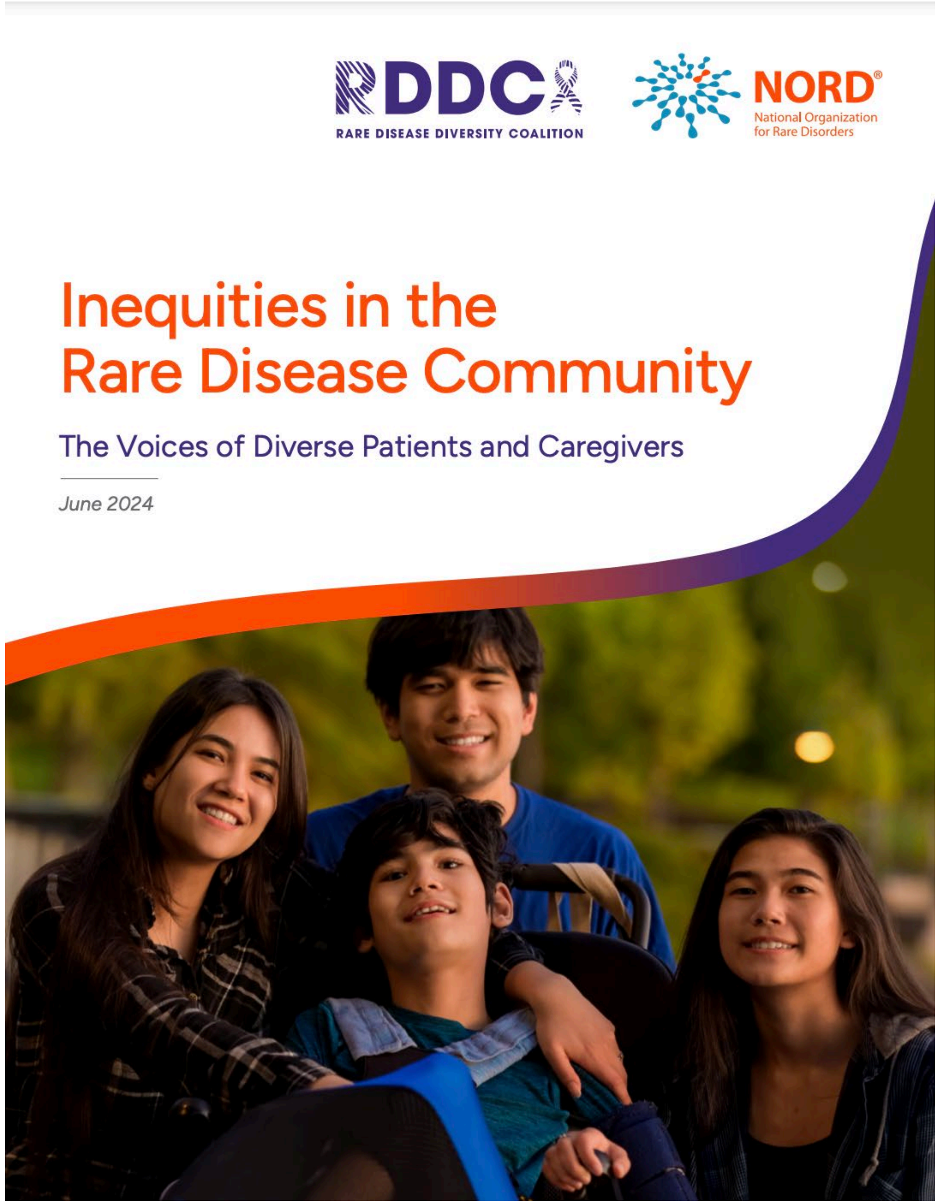
For historically marginalized persons with a rare disease, these challenges are compounded by institutionalized disparities that have resulted in underrepresentation in studies and clinical research trials, leading to a lack of understanding about effective treatments.

ABOUT RDDC

Black Women's Health Imperative (BWHI) launched the Rare Disease Diversity Coalition™ (RDDC™) to address the extraordinary challenges faced by underserved populations with rare diseases.

The Coalition brings together rare disease experts, patients, caregivers, health care professionals, community advocates, and industry leaders to develop evidence-based solutions that reduce the disproportionate burden of rare diseases on marginalized populations.

RDDC
THE RARE DISEASE DIVERSITY COALITION



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

SCOPE OF PROJECT

Uncovering gaps and barriers experienced by medically underrepresented populations (patients and caregivers).

Sponsor: Rare Disease Diversity Coalition (RDDC)

Team: Patient, Provider, Caregiver Group

Partner: NORD (National Organization for Rare Disorders)

Released: August 2024

BLACK WOMEN'S
HEALTH IMPERATIVE

RDDC™
RARE DISEASE DIVERSITY COALITION

NORD®
National Organization
for Rare Disorders

NORD®
National Organization for Rare Disorders

SURVEY OBJECTIVES

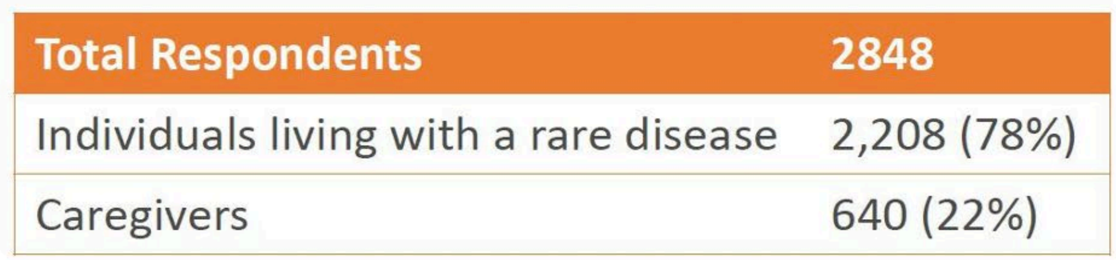
- Quantify the actual, and perceived, barriers and gaps faced by historically underrepresented rare disease patients in accessing diagnosis, clinical care, and treatment.
- Identify how these gaps and barriers may be different across various population groups.
- Facilitate a more profound comprehension of these gaps and barriers.
- Inform the development of recommendations to address the barriers to care among marginalized rare disease patients.

NORD®
National Organization for Rare Disorders

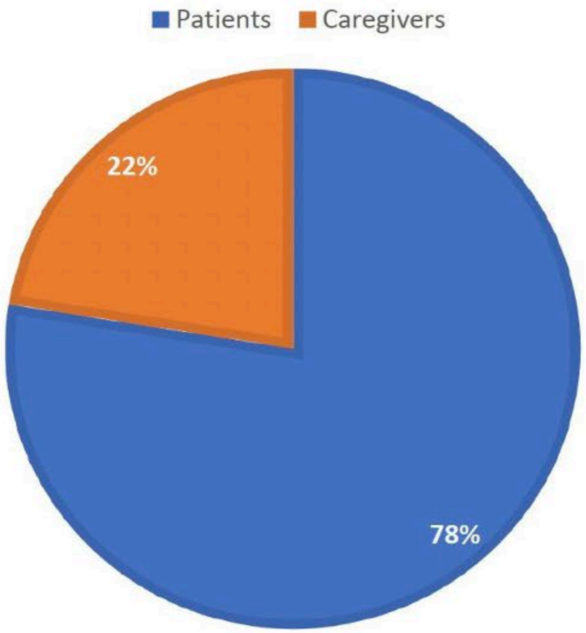
RECRUITMENT STRATEGIES

- Promotion in English and Spanish
- E-mail distribution to RDDC/NORD/Working Group networks of tens of thousands
- Ethnic based advocacy groups
- Postcard promotion at exhibits at health conferences and grassroots community health fairs nationwide
- Promotion to 100,000s via multi-stakeholder social media channels
- Paid Google ads

SURVEY RESULTS – SUMMARY



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



SURVEY RESULTS – SUMMARY

NORD®
National Organization for Rare Disorders

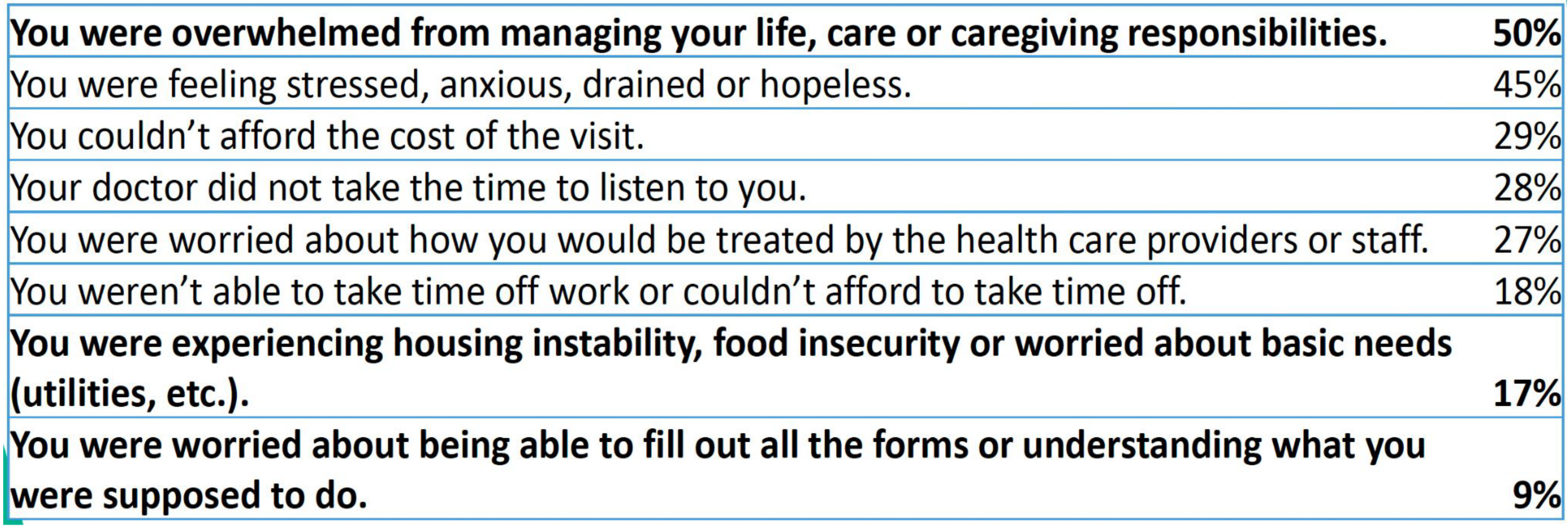
Underrepresented Respondents

	Count (%)
BIPOC	448 (18%)
Hispanic Ethnicity	204 (8%)
LGBTQ+	329 (14%)
Household income below poverty level	433 (17%)
Rural Residence	270 (13%)
Total Unique Individuals	1259 (48%)

SURVEY AUDIENCES & DEFINITIONS

- BIPOC – Individuals that identify with at least one of the following: Black, Indigenous or a Person of Color.
- Hispanic Ethnicity – Individuals that identify with Hispanic, LatinX, or Latine ethnicity
- LGBTQ+ – Responded to questions about sexual orientation and gender identity indicating that they were at least one of: gay, lesbian, bisexual, transgender, non-binary or other self-reported terms used to indicate LGBTQ+ status
- Federal Poverty Level – Determined based on reported income range and household size based on 100% of the 2023 Federal Poverty Level
- Rural Residence - Determined based on reported zip code of residence and then the Rural-Urban Continuum Codes were applied.

SURVEY GENERAL RESULTS



You were overwhelmed from managing your life, care or caregiving responsibilities.	**50%**
You were feeling stressed, anxious, drained or hopeless.	45%
You couldn't afford the cost of the visit.	29%
Your doctor did not take the time to listen to you.	28%
You were worried about how you would be treated by the health care providers or staff.	27%
You weren't able to take time off work or couldn't afford to take time off.	18%
You were experiencing housing instability, food insecurity or worried about basic needs (utilities, etc.).	**17%**
You were worried about being able to fill out all the forms or understanding what you were supposed to do.	**9%**

Goal: to collect information from a diverse sample of individuals with a rare disease and those who care for them, to better understand the experiences of individuals living with a rare disease and investigate whether ***differences are observed across populations.***

SURVEY RESULTS – FINANCIAL BARRIERS

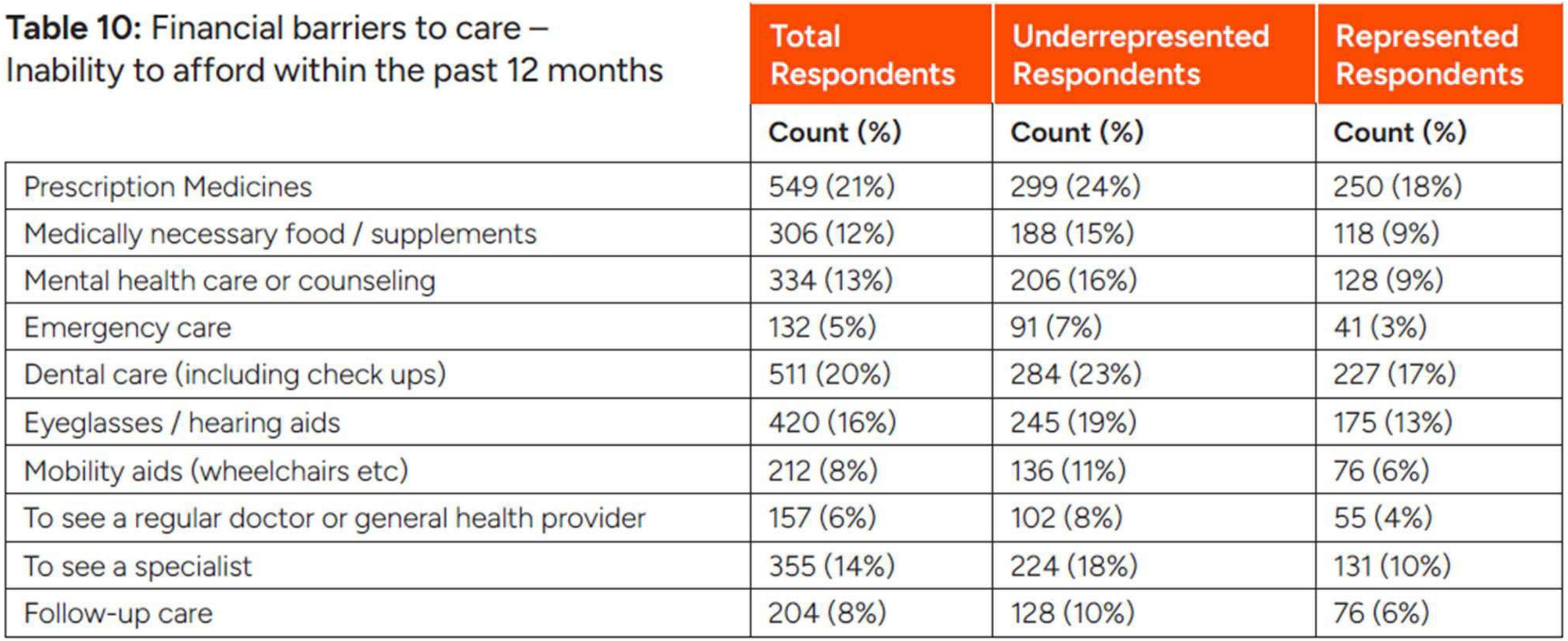
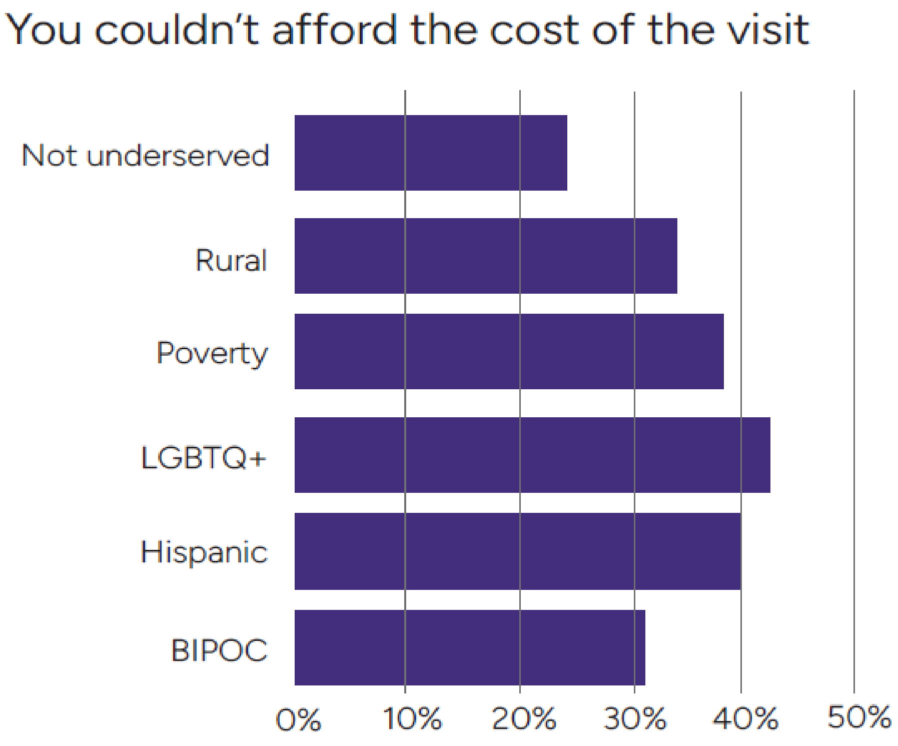


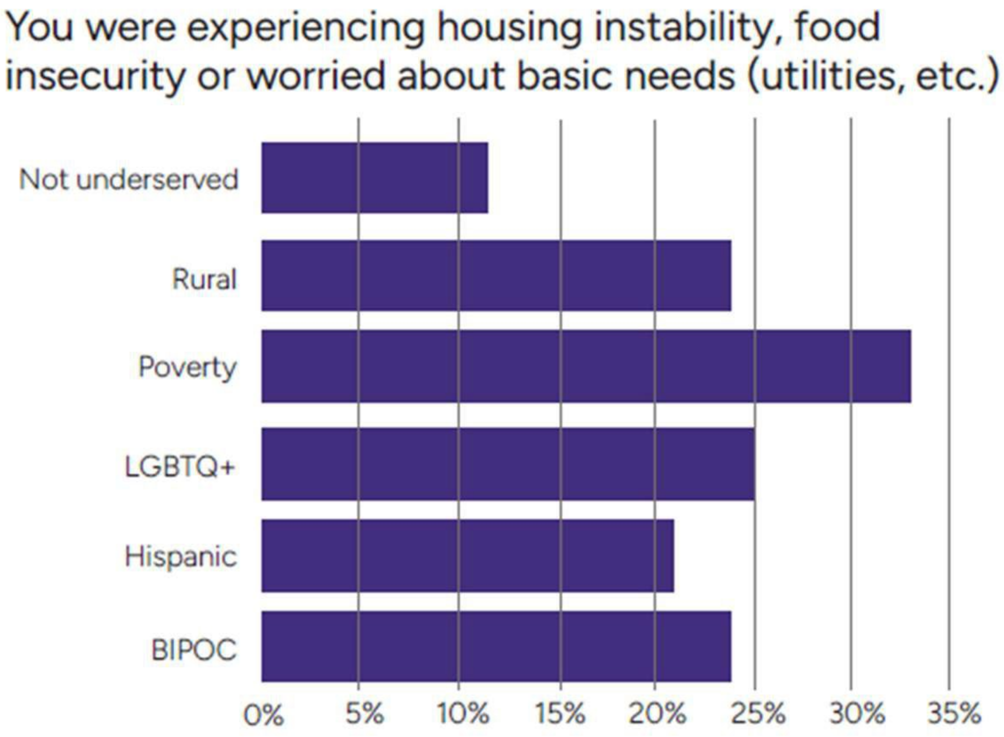
Table 10: Financial barriers to care – Inability to afford within the past 12 months

	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%)

SURVEY RESULTS – RARE DISEASE WARRIOR HIGHLIGHTS



SURVEY RESULTS – RARE DISEASE WARRIOR HIGHLIGHTS



SURVEY RESULTS – CAREGIVER HIGHLIGHTS

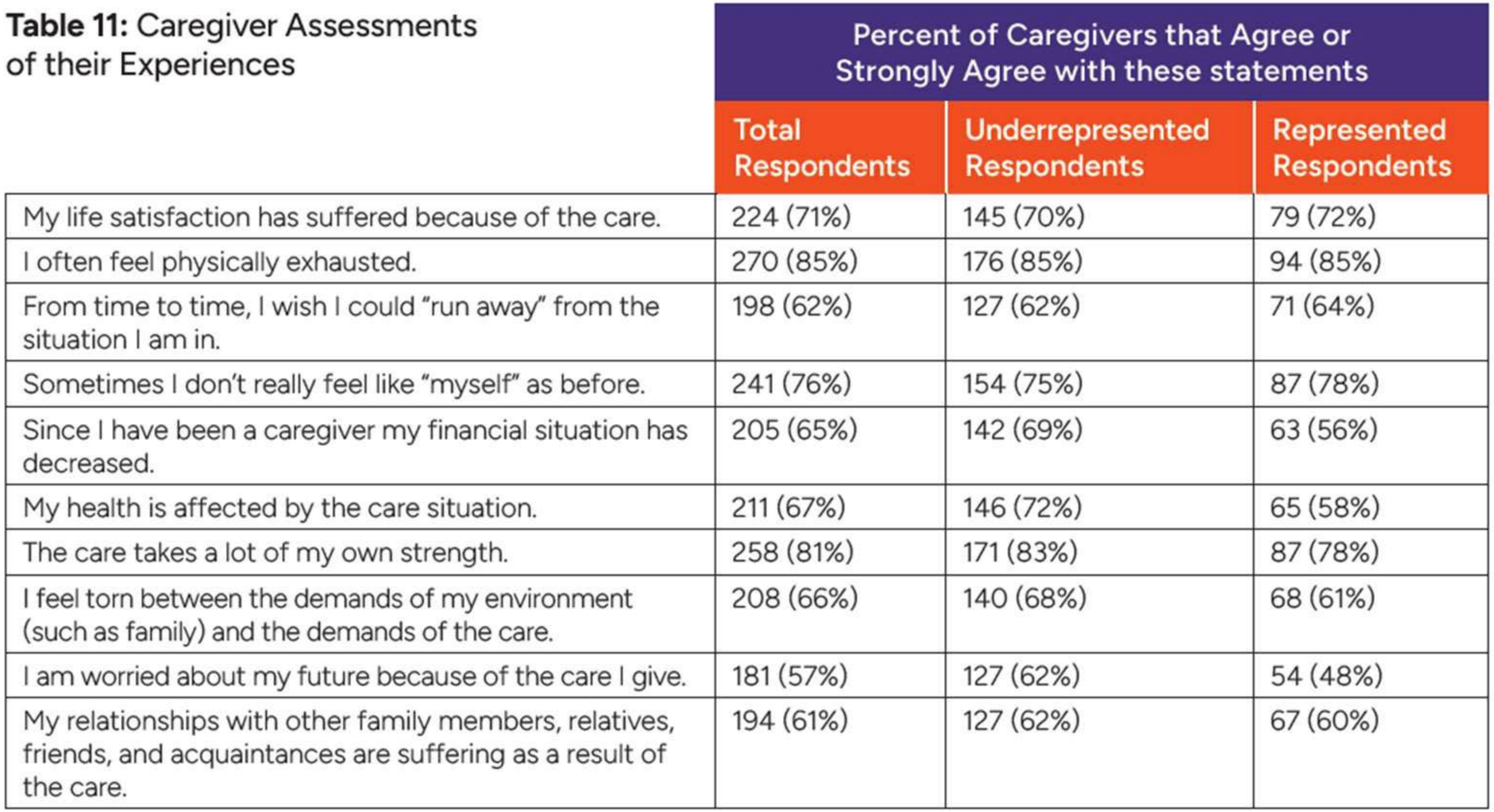
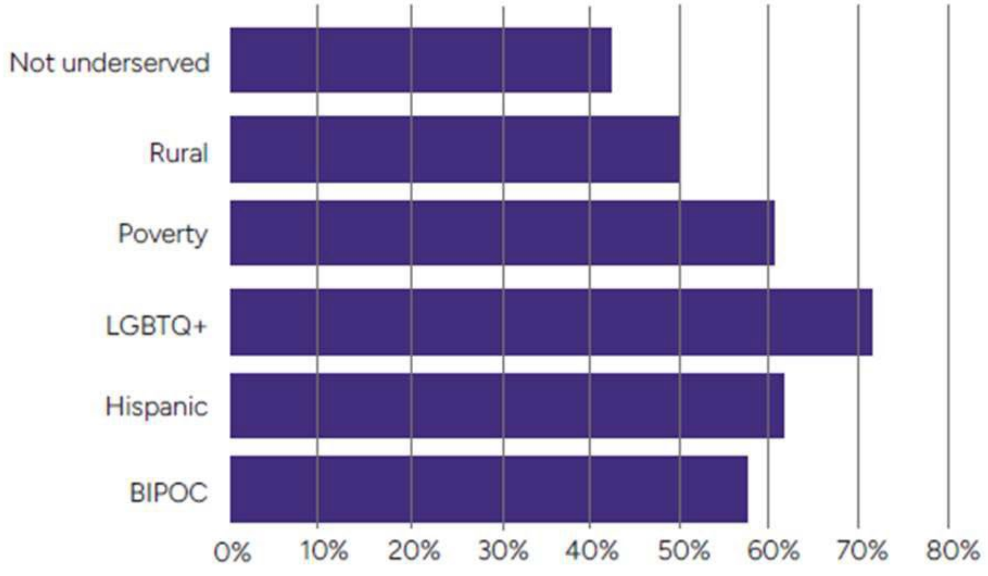


Table 11: Caregiver Assessments of their Experiences

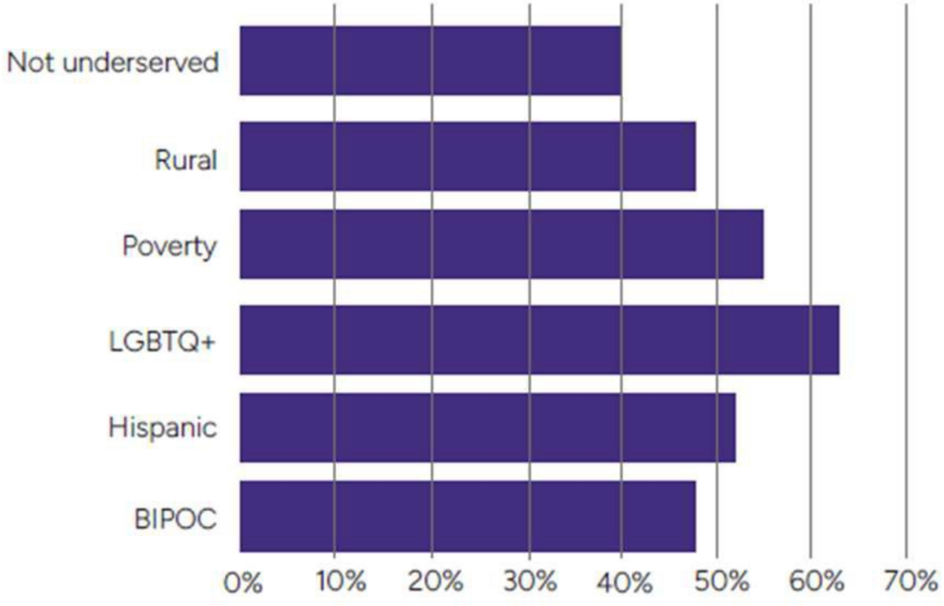
	Percent of Caregivers that Agree or Strongly Agree with these statements		
	Total Respondents	Underrepresented Respondents	Represented Respondents
My life satisfaction has suffered because of the care.	224 (71%)	145 (70%)	79 (72%)
I often feel physically exhausted.	270 (85%)	176 (85%)	94 (85%)
From time 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%)

SURVEY RESULTS – CAREGIVER HIGHLIGHTS

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



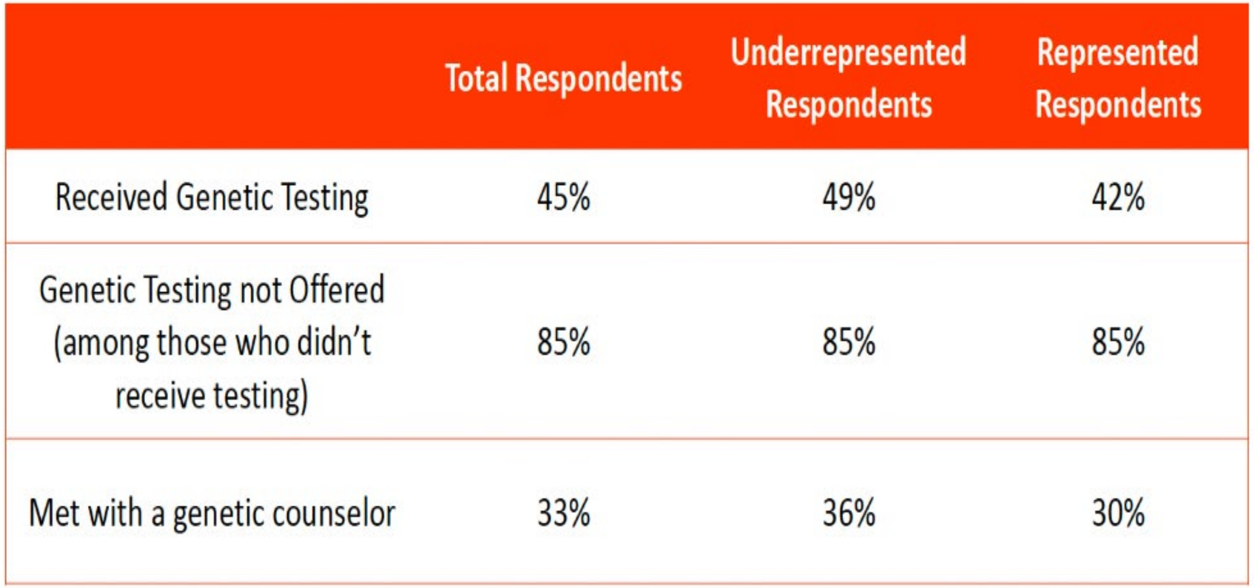
You were feeling stressed, anxious, drained or hopeless



ADDITIONAL FINDINGS & OPPORTUNITIES

The Diagnostic Journey: Improving Access to Genetic Testing

72% of rare diseases are genetic in origin but fewer than half of respondents have received genetic testing and of those who did not receive testing, 85% were never offered.



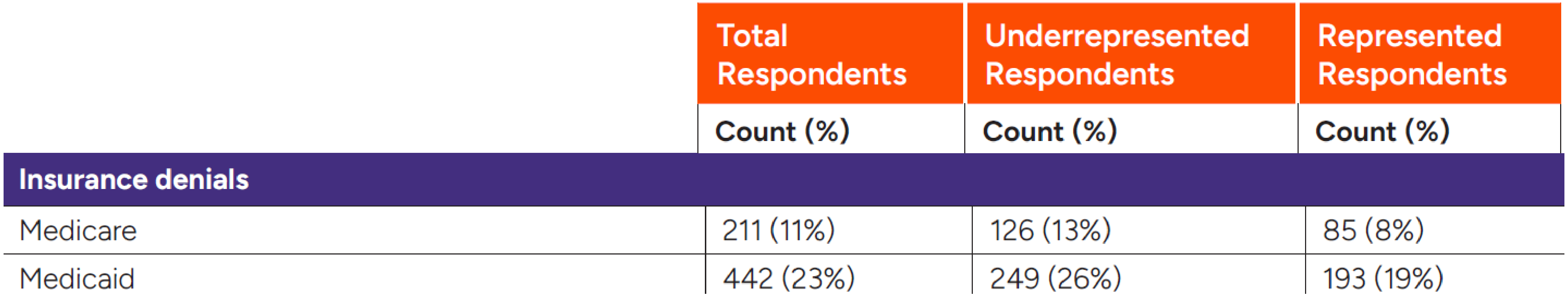
	Total Respondents	Underrepresented Respondents	Represented Respondents
Received Genetic Testing	45%	49%	42%
Genetic Testing not Offered (among those who didn't receive testing)	85%	85%	85%
Met with a genetic counselor	33%	36%	30%

ADDITIONAL FINDINGS & OPPORTUNITIES

Navigating Health Insurance: Addressing Insurance Denials

93% of all respondents had health insurance (49% Private Insurance, 35% Medicare, 16% Medicaid)

Medicare denials were reported by 11% of respondents and Medicaid denials were reported by 23% of respondents. **The proportion of denials was higher in under-represented communities.**



	Total Respondents	Underrepresented Respondents	Represented Respondents
	Count (%)	**Count (%)**	**Count (%)**
Insurance denials			
Medicare	211 (11%)	126 (13%)	85 (8%)
Medicaid	442 (23%)	249 (26%)	193 (19%)

MEDICATION ACCESS SOLUTIONS

- **Expand Insurance Coverage:** Advocate for policy changes to cover orphan drugs, genetic testing, and specialty medications under Medicaid and private insurance.
- **Improve Provider Training:** Increase cultural competency in prescribing practices and ensure rare disease education includes diverse patient populations.
- **Strengthen Patient Assistance Programs (PAPs):** Expand financial assistance and community-based medication distribution programs to help patients afford and access necessary treatments.
- **Increase Diversity in Clinical Trials:** Ensure drug development includes patients from diverse racial and socioeconomic backgrounds to improve equity in treatment outcomes.

Question and Answer

Please submit your questions using the Q&A feature at the bottom of your screen

PQA Announcements

Upcoming PQA Events

25 PQA

ANNUAL MEETING

TAMPA, FL | MAY 19-21, 2025

Upcoming PQA Events

Medication Use Quality:
A PQA Live CE Event

Monday, May 19, 2025

8:00 a.m.-12:00 p.m. ET

Tampa, Fla.

Pharmacists earn 0.4 CEU

2024 PQA Stakeholder Advisory Meetings

PQA STAKEHOLDER ADVISORY MEETINGS

First Quarter SAM: March 12, 2025, 1-2 pm ET

- Summary of the CMS Proposed Rule (CMS-4208-P) and 2026 Advance Notice
- QuIRC project updates on:
    - PQA's project to evaluate the impact of the Medicare cost cap for insulin on patient persistence
    - The second phase of PQA's work to improve the quality of oral anticancer medication use
    - PQA's pilot to evaluate the effectiveness of pharmacies in improving adult immunization rates through value-based arrangements with payers.
- Highlights from the PQA Health Equity Technical Expert Panel (TEP) report
- Recent PQA publications focused on medication therapy management

Second Quarter SAM: June 11, 2025, 1-2 pm ET
Third Quarter SAM: August 13, 2025, 1-2 pm ET
Fourth Quarter SAM: October 22, 2025, 1-2 pm ET

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