RARE DISEASE WEEK ON CAPITOL HILL 2021

Diversity Roundtable Discussions

Abstract

On July 22, 2021, the EveryLife Foundation for Rare Diseases held its tenth annual Rare Disease Week on Capitol Hill. EveryLife was joined by distinguished leaders from 16 organizations who specialize in minority health policy and rare disease and representative staff members from the Congressional Black Caucus, Congressional Hispanic Caucus, Congressional Asian Pacific American Caucus, and the Congressional LGBTQ+ Equality Caucus.

Priscilla Rodriguez, Diversity Inclusion Advocacy Fellow

EveryLife Foundation Roundtable Organizer*: Adrian Palau-Tejeda*

INTRODUCTION

On July 22, 2021, Rare Disease Legislative Advocates, a program of the EveryLife Foundation for Rare Diseases, held its tenth annual Rare Disease Week on Capitol Hill. This event served as an opportunity to influence legislative change for the estimated 30 million rare disease patients living in the United States. EveryLife was joined by distinguished leaders from 16 organizations who specialize in minority health policy and rare diseases and representative staff members from the Congressional Black Caucus, Congressional Hispanic Caucus, Congressional Asian Pacific American Caucus, and the Congressional LGBTQ+ Equality Caucus. The goal of the Foundation and its partners was to hold distinct and concurrent conversations on issues impacting the health outcomes of people from underserved and underrepresented communities who are also living with a rare disease. Lessons learned from the roundtables will assist stakeholders in ensuring that meaningful steps are taken to meet the health equity priorities of the diverse rare disease community.

BACKGROUND

In the United States, rare diseases are defined as any disease that has a patient population below 200,000 individuals. There are more than 7,000 rare diseases impacting millions of Americans (Yang et al., 2022). The prevalence of diseases that are considered to be rare span from those such as cystic fibrosis, sickle cell disease, and some forms of muscular dystrophy where patient populations may include 5,000-10,000 people living in the U.S., to diseases considered to be 'ultra-rare' in which there may be fewer than 5 or 10 people living in the U.S. Individuals and families living with rare diseases and conditions face a variety of obstacles including timely and accurate diagnosis, lack of effective therapies and cures, and prohibitive costs for life-sustaining interventions. In rare and ultra-rare diseases, advocacy efforts and programs and concerted efforts of stakeholders are crucial to securing funding for inclusive research, furthering medical product development, and improving drug approval pathways at the FDA.

Yang et al.'s (2022) *The National Economic Burden of Rare Disease in the United States in 2019* found "the estimated total economic burden of 379 rare diseases with a prevalence of 15.5 million people in 2019 was $966 billion, including a direct medical cost of $418 billion and an additional $548 billion in indirect and nonmedical cost." The diagnostic odyssey for underserved communities is further complicated by social determinants of health and higher economic constraints, which leads to worse health outcomes. The September 2021 U.S. Government Accountability Office (GAO) *Racial and Ethnic Health Disparities* report found that "Black women experienced 59.3 deaths per 100,000 live births compared to only 19.7 deaths per 100,000 live births for White women living in the same counties." This is only one example of the inequities and injustices faced by underserved communities in this country. The Rare Disease Week 2021 Diversity Roundtables provided a comprehensive overview of the challenges faced by the diverse rare disease community and proposed policy solutions to address the gaps that continue to exist.

ROUNDTABLE METHOD

A total of four virtual diversity roundtable discussions were held on the topics of Access, Representation, Clinical Trials, and Newborn Screening and Diagnostics. Key opinion leaders were invited to share their perspectives on the many challenges faced by underserved populations during the discussions. Patient-advocates shared their stories about their diagnostic odysseys and the challenges still to come. A staff representative from the Congressional Black Caucus, Congressional Hispanic Caucus, Congressional Asian Pacific American Caucus, and the Congressional LGBTQ+ Equality Caucus were included in the conversations.

CURRENT STATUS OF RARE DISEASE POLICY PRIORITIES

First, it is necessary to provide context on the current rare disease policy priorities and status as they relate to the roundtable discussions. Patients face numerous barriers that prevent them from accessing care and/or treatments that are capable of enhancing their health. This includes barriers to out-of-state care which limits traveling for treatment and diagnostics, restrictions to appropriate clinical providers, and complexities in proper documentation. Further issues include the inter-play of patient representation in the Medicaid Drug Utilization Review (DUR/P&T Committee) process and workplace discrimination for rare conditions as a result of ERISA/self-funded plans ability to zero in on costs for treatments for specific diseases.

Value assessments and the implementation of value-based payment arrangements that are anchored in diverse, patient-centered metrics remain critical to ensure that approved therapies are improving health outcomes. Coverage of genetic testing services, telehealth, and revised clinical trial approaches are major policy priorities for the underserved rare disease community.

Newborn screening is a proven tool to diagnose and provide early intervention to the rare disease population. Through this public health program, newborns are screened at birth for a wide range of conditions, including racial and ethnically prevalent disorders like sickle cell disease. Through the implementation of this program, there has been some progress in closing the gap in detection of rare conditions that are screened for within underserved rare disease patients, but more can and must be done to make diagnostics, treatments, and cures more equitable.

Cures 2.0 legislation, introduced by Represetatives Diana DeGette (CO) and Fred Upton (MI) in 2021, builds on the 21st Century Cures Act. This legislation aims to improve FDA-CMS communication regarding transformative new therapies, increase the use of real-world evidence in regulatory decision making, and expand access to genetic testing. In addition, in May of 2022 the Department of Health and Human Services established the Advanced Research Projects Agency for Health (ARPA-H) within the National Institutes of Health (NIH). While ARPA-H implementation is still nascent, it serves as another important opportunity for the underserved rare

disease community to be considered in future programming.

The seventh reauthorization of the Presciption Drug Fee User Act (PDUFA VII) introduced in 2022 also includes a commitment to enhancing clinical trial diversity within therapeutic development.

ROUNDTABLE SECTION: ACCESS

Moderator: Jamie Sullivan, EveryLife Foundation for Rare Diseases
Panel:
Scout, LGBT Cancer Network
Lisa Bonebrake, Alport Syndrome Foundation
Carmen Camacho, HPS Network
Pamela Price, Balm in Gilead
Isabel Rangel, Congressional Hispanic Caucus

The Access Roundtable began by addressing the unique challenges faced by underserved rare disease communities through the panelists' perspectives on community specific risks and their contribution to worsening health outcomes. Panelists discussed **diagnostic and clinical appointment wait times, coverage issues, poor patient-provider relationships,** and external factors that significantly impact the diagnostic process.

A rare disease diagnosis is often measured in the span of years and may involve interacting with dozens of physicans and specialists. One panelist discussed the challenges associated with rare cancers. HIV positive patients are at a greater risk of developing rare cancers due to the inability to fight off an HPV infection. Patients at high risk of HIV, such as the LGBTQ+ community, often struggle to access adequate care due to their reluctance to share health information, which may result in a misdiagnosis or untreated infections. Systemic distrust in the healthcare system remains a principal barrier for communities seeking diagnostics and treatment for rare disorders and conditions.

Miscommunication and language barriers also contribute to the disconnect between health providers and the patients they serve. It is critical that patients understand whether a condition is genetic or has inherited characteristics to prevent disease or better manage their health. Another panelist linked her own experience of **miscommunication with her health care provider.** Physicians failed to inform her of the risks associated with her x-linked rare disorder, exacerbating challenges as her child was also diagnosed. Panelists spoke of the need for better communication as well as greater access to and use of services and resources.

Genetic counseling, testing services, and cultural compentency training were all deemed imperative by the panel. One panelist drew an example from a privately funded, but free-for-patients genetic testing program that relied on the recommendations of physicians. Shortcomings were observed when additional testing was not recommended, only for genetic conditions or carrier status to later be discovered.

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Telehealth and geographic location barriers were other topics brought up in the discussion. The panelists underscored the importance of policy solutions for rare and underserved communities that are focused on increasing at-home doctors' visits and improving access to care across state lines. Medicaid expansion and cross-state provider mobility remain paramount for coverage and a principal step in establishing health equity.

Due to historical effects of conflict and federal neglect, geographically isolated regions, such as Puerto Rico, face additional access barriers, which force the population to seek medical treatment outside of the island. *The National Economic Burden of Rare Disease in the United States in 2019* (Yang et al., 2022) found the mean direct costs for metabolic disorders, like Hermansky-Pudlak Syndrome, to be $39,432 per year. As one panelist noted, expanding coverage and access will not equal utilization. Barriers still exist beyond coverage, inter-state travel, and economic costs.

ROUNDTABLE SECTION: REPRESENTATION

Moderator: Adrian Palau-Tejeda, EveryLife Foundation for Rare Diseases
Panel:
J. Maurice McCants, Human Rights Campaign
Tammy Boyd, Black Women's Health Imperative
Gustavo Paredes, Hispanic Institute
Alpa Khushalani, Parent Project Muscular Dystrophy
Shawn Gaylord, LGBT Equality Caucus

The COVID-19 pandemic brought into creation the Rare Disease Diversity Coalition (RDDC), a group dedicated to understanding and addressing the needs of the diverse rare disease population. Though a noteworthy accomplishment, panelists drew on the importance of greater advocacy efforts and the need to hold elected officials accountable to drive policy change and increase diverse representation. The **need for meaningful outreach and engagement with underreprented populations** was mentioned repeatedly.

Cultural barriers was one issue identified by the panelists. Both patient advocates and diverse community leaders focused on the **need of language-based policy solutions that call for specific language that focuses on the individual rather than the disease.** One patient advocate discussed their own personal experience with stigmatization and how labels and definitions can influence one's ability to seek care. Representation and empowerment of underserved patients begins with the terminology used to define demographics and characteristics.

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The need to reform the medical student to doctor pipleline was also mentioned, citing that out of the 30,000 medical students in the nation, only 1,600 are Black. Within the current system, doctors leave medical school unprepared to collaborate with patients of color. Another panelist shared their experience working to advance national Hispanic community efforts. Panelists argued that federal programs have

historically failed to intentionally reach underserved communities, and previous census efforts failed to accurately characterize diverse individuals. Despite the more than 44 million Spanish speakers in the United States, the need for language-sensitive information, Spanish-speaking providers, and engagement with Spanish language media is critical.

Roundtable panelists pointed to the Accelerated Approval Pathway within the FDA as a key concern for underserved community representation. Challenges include the ability of insurers to deny coverage due to limited testing for an entire subpopulation or determine that a treatment is cost prohibitive or unnecessary. Referencing the Better Empowerment Now to Enhance Framework and Improve Treatments (BENEFIT) Act, patient advocates drew on **the importance of increasing and ensuring greater involvement of the patient voice in the drug development process.**

Representation in legislative advocacy remains an important goal for diverse communities. Panelists emphasized the **need for representation in both advocacy and the Congressional body.** It is crucial that advocacy organizations reach underrepresented individuals and groups, so their stories are heard on a broader scale and reflected in legislation and policy. While Congress has become more diverse, further work is needed to ensure that every community is reflected in the Congressional leadership pipeline. The constant call for representation is a top priority for the diverse rare disease community, and it is evident that we must continue to hold our local, state, and federal legislators accountable.

ROUNDTABLE SECTION: CLINICAL TRIALS

Moderator: Annie Kennedy, EveryLife Foundation for Rare Diseases
Panel:
Lauren Lee, Nepchure International
Barbara Bierer, Multi-regional Clinical Trial Network
Adrienne Shapiro, Sick Cells
Teneasha Washington, Rare-X
Mia Keeys, Office of Representative Robin Kelly

Panelists referenced their own personal experience and the need for diversity in trial design and recruitment. A case of poor representation in clinical trials was described in the Journal of Clinical Oncology. From 2009 to 2015, only 3% of participants in prostate cancer clinical trials were black, despite the fact that black men are twice as likely to develop prostate cancer as white men. The 2017 Food and Drug Administration's (FDA) Global Participation in Clinical Trials Report showed that Black or African American participation in general oncology clinical trials was only 2.75% from 2015-2017. A stepping stone to increase diversity in clinical trials is the establishment of partnerships with trusted community leaders. Utilizing trusted messengers and community influencers enables sponsors to engage in meaningful outreach with diverse communities.

Inaccurate and non-existent data collection further exacerbates the safety and effectiveness gap for different subpopulations. Due to the historical relationship between researchers and underserved populations, especially racial

and ethnic minorities, individuals today are less likely to participate or be represented in research trials and studies. Despite the use of different data collection platforms by major government agencies, novel projects like the Oral History Projects of Columbia University or the National Institutes of Health's All of US program offer hope for diverse inclusion in trials and data for rare diseases.

Panelists brought to light the cultural and historical injustices that have led to poor treatment, generational trauma, and lack of trust in the healthcare system. Throughout the discussion, speakers emphasized that in order to combat the lack of participation of underserved communities in clinical trials, it's crucial to **listen and understand the issues and concerns facing these groups.** Demographic subgroups, geographic location, language barriers, and socioeconomic status of all participants in a clinical trial needs to be considered. Only 19% of clinical trials require individuals to read, write, and speak English to participate. More work needs to be done to improve health literacy and ensure equity demands are met, given the ongoing social battles and changes in our nation and across the globe.

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ROUNDTABLE SECTION: NEWBORN SCREENING AND DIAGNOSTICS

Moderator: Dylan Simon, EveryLife Foundation for Rare Diseases
Panel:
Naomi Louie, Asian Health Coalition
Marissa Penrod, Team Joseph
Gary Gibson, Sickle Cell Association of America
Natasha Bonhomme, Expecting Health
Anisha Assim, Asian and Pacific American Caucus

Newborn screening remains one of the most pivotal health equity programs within the United States. While all states may not screen for the same conditions, newborn screening has yielded improved health outcomes for families across the nation. However, conversation focused around the barriers and inequities that exist within diagnostics and newborn screening. Participants **called for enactment of legislation such as the Health Equity and Accountability Act** that would increase access to health care for underserved communities.

Systemic racism, inadequate information and education, cultural incompetence, and other factors negatively contribute to the diagnostic and follow-up process. For underserved communities, disparities in care start in the pre-diagnosis stage. In addition to the stress caused by external circumstances, the newborn screening process can be overwhelming and confusing**. Health literate information and telehealth services** provide an opportunity for diverse populations to address the complexities and challenges that are a part of the process. In programs like family assistance, we can learn about a family's situation and provide them with the

resources and assistance they need to succeed.

Physician refusal to assess risk for certain conditions based on demographics leads to babies being misdiagnosed or undiagnosed. Panelists also stood against a non-uniformed nature of testing, citing the goal should be to minimize the risk of someone missing potential care.

A challenge raised during the discussion was the long wait between the time babies are born and the time the screening results are released. A speaker noted that nearly ten percent of patients are difficult to reach after screening results are made available. When it comes to rare diseases like sickle cell, patients may not realize that they are trait carriers, may deny the results, or may move and forget to give their health institutions updated information.

Geographic location and lack of communication were deemed problems in diagnosis and follow-up care for newborns. Due to the lack of specialists in certain areas, traveling to specialty centers may be daunting for many families. Panelists also found an issue in communication between physician offices and specialists. Lack of data collection and segregation of data contribute to the circulation of insufficient diagnosis and treatment. Panelists pushed for **greater accountability and recognition of institutions based on performance.**

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ANALYSIS

The GAO (2021) report on *Racial and Ethnic Health Disparities* found that racial and ethnic minorities are at greater risk of dying from preventable causes and chronic conditions than other Americans. Common themes that emerged from the roundtable discussions were: 1) understanding the systemic issues and stressors behind poorer health outcomes for underserved communities; 2) data under-representation; and 3) the need to have the patient voice heard.

Partnerships with community leaders are important for reaching the most marginalized. Trusted community partners serve as important collaborators in outreach due to their shared experiences and constant engagement with overlooked populations.

With significant barriers to screening, testing and follow-up care, policies should focus on increasing access to genetic testing services and counselors, coverage across state lines, and making sure provided resources are utilized. Current coverage for recommended genetic testing, especially whole genome and whole exome sequencing, is lacking and requirements for obtaining coverage are lengthy and complex. This is partially driven by improper cultural competency training, lack of information from physicians, and outdated screening diagnostic methods such as race based recommendations.

Improper data collection and utilization of diverse population demographics is evident in the rare disease space. The 2021 GAO

report called for new GAO policy recommendations including "complete reporting of race and ethnicity information by states, jurisdictions, and federal health systems and the routine analysis of health outcomes using data that can help the federal government better understand existing health disparities and take actions to promote health equity."

The Rare Disease Week on Capitol Hill 2021 panelists repeatedly pointed at the need for greater reporting data to come from research and governmental institutions working towards building meaningful relationships with communities that have historically been ignored. The GAO (2021) report further noted that "the consideration of racial and ethnic disparities within federal government programs can help agencies determine how their goals can be operationalized to advance health equity." The policy perspectives gathered from the roundtables were firmly rooted in the empowerment and involvement of patients in the data collection process as both subjects and active partners.

CONCLUSION

Policies must aim to address inappropriate healthcare management practices, provider-patient communication, inclusion of diverse patients in clinical trials, and inaccurate data collection platforms. When discussing review processes and decision making in all aspects related to rare disease policymaking, drug development, and regulatory decisions, diverse patient representation must be included. The rare disease community works tirelessly to close the innovation gap for the 93% of rare diseases that have no FDA-approved treatment, eliminate the diagnostic odyssey for rare disease patients, improve the regulatory process and advance regulatory science for rare diseases therapies, and ensure patients have access to safe and efficacious therapies and cures at the earliest moment possible. As rare disease advocates, government officials, and health sector leaders, it is our responsibility to take all necessary steps to guarantee that every voice is heard, and immediate action is taken.

REFERENCES

1. Agency for Healthcare Research & Quality. (2012). Improving patient safety systems for patientd with limited english proficiency. Retrieved from https://www.ahrq.gov/sites/default/files/publications/files/lepguide.pdf
2. Government Accountability Office. (2021). Racial and ethinic health disparities. Retrieved from https://www.gao.gov/assets/720/716930.pdf
3. Shenoy, D., Packianathan, S., Chen, M. A., & Vijayakumar, S. (2016). Do African-American men need separate prostate cancer screening guidelines? *BMC Urology, 16*(19), 1-6. Doi: 10.1186/s12894-016-0137-7
4. Stier, E. (2008). Human Papillomavirus related diseases in HIV-infected individuals. *Current Opinion in Oncology, 20*(5), 541-546. Doi: 10.1097/CCO.0b013e3283094ed8

5. U.S. Food & Drug Administration. (2017). 2015-2019 Drug trials snapshots summary report. Retrieved from https://www.fda.gov/media/143592/download
6. Yang, G., Inna, C., Pariser, A., Oehrlein, E., Sullivan, J., & Kennedy, A. (2022). The national economic burden of rare disease in the United States in 2019. *Orphanet Journal of Rare Diseases, 17*(163), 1-11. https://doi.org/10.1186/s13023-022-02299-5

AUTHOR

Priscilla Rodriguez is the Diversity Inclusion Advocacy Fellow at the EveryLife Foundation for Rare Diseases. In her work at EveryLife, Priscilla ensures an intentional, effective, and sustainable DEI focus in all Foundation programming and across its partner and peer organizations. She is a master's degree candidate (Dec. 2022) in public health at The George Washington University - Milken Institute School of Public Health.