



RARE DISEASE DIVERSITY COALITION

Health Equity for People Living with PNH: Creating a Leveled Playing Field



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About the Rare Disease Diversity Coalition



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 strategies and organizations to 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 diseases. 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. Learn more at rare diseasediversity.org.



About Links2Equity



Links2Equity is a mission-driven patient advocacy consulting firm that gives patients and families, advocacy professionals, and other healthcare stakeholders a voice. With over 25 years of experience in patient advocacy and government affairs, Links2Equity aims to empower healthcare stakeholders to shape a more equitable healthcare system. The firm provides strategic consulting services to support patient

advocacy and health equity efforts for all disease states and underrepresented communities. By uniting healthcare stakeholders and underrepresented patient populations, Links2Equity builds strong relationships and fosters collaborative dialogue to deliver a better patient experience and minimize health disparities. Learn more at Links2Equity.com.

Executive Summary

This white paper was developed to achieve greater equity for patients living with Paroxysmal Nocturnal Hemoglobinuria (PNH) with recommendations to improve equitable access to care, reduce financial barriers, and enhance provider education.

The Rare Disease Diversity Coalition (RDDC) formed the PNH Health Equity Steering Committee, bringing together patient advocates, healthcare professionals, caregivers, and key stakeholders to explore inequities driven by socioeconomic status, systemic biases, and geographical barriers. The paper highlights barriers to care, particularly for minoritized and marginalized groups, including Black and Indigenous People of Color (BIPOC); Lesbian, Gay, Bisexual, Transgender, Queer and/or questioning, Intersex, Asexual, and other diverse identities (LGBTQIA+); residents of rural areas; people with incomes below the federal poverty level; and immigrants with limited English proficiency who experience greater barriers to timely diagnosis, treatment, and ongoing care.

Key recommendations:

Enhancing the patient and care giver

Improving access to FDA-approved treatment

Reducing financial barriers

Integrating cultural competency

Gender and gender diversity in care

Expanding telehealth services

Promoting diversity in clinical research

These efforts aim to reduce disparities and create more equitable care for all PNH patients, addressing broader inequities in the rare disease community.

Introduction

Rare or “orphan” diseases profoundly impact the human experience. Over 400 million individuals globally are living with a rare disease—this includes 30 million people in the United States, who represent over 10,000 distinct diseases and conditions.¹

Within the rare disease community, some individuals and families live with “ultra-rare” conditions. For these patients and caregivers, the challenges of living with a rare disease, getting an appropriate diagnosis, and finding appropriate care are amplified. The ultra-rare disease Paroxysmal Nocturnal Hemoglobinuria (PNH) is a blood disorder affecting approximately 0.5 to 1.5 cases per million people. For a condition where fewer than two people will have PNH for every one million patients, its rare nature leads to challenges in obtaining timely diagnoses, specialized care, and effective treatments.²

The ultra-rare disease Paroxysmal Nocturnal Hemoglobinuria (PNH) is a blood disorder affecting approximately 0.5 to 1.5 cases per million people.

Barriers to Better Health for Rare Disease

People and families who live with rare diseases often face a difficult and frustrating journey to find treatment and support. Rare disease patients and caregivers experience significant challenges, which amplify existing inequities in the treatment of these diseases.

Challenges facing rare disease patients included:



Delayed diagnosis and length of time to diagnosis

Conditions often take years to diagnose due to their complexity, and patients may not have access to knowledgeable medical professionals who can accurately diagnose their condition.³



Limited availability of treatments

Many rare diseases lack FDA-approved treatments, leaving people without effective treatment options and resulting in high medical costs and poor patient outcomes.⁴

Fractured access to health insurance

¹ <https://rarediseases.org/rare-diseases/>, <https://globalgenes.org/rare-disease-facts/>

² <https://rarediseases.org/rare-diseases/paroxysmal-nocturnal-hemoglobinuria/> (See the “Affected Populations” section)

³ https://ncats.nih.gov/sites/default/files/NCATS_RareDiseasesFactSheet.pdf

⁴ *Id.*



Disparities in health insurance systems lead to unequal access to medications, particularly in rural and lower socioeconomic regions, where patients may experience longer approval times or denials for lifesaving treatments.⁵



Financial strain and toxicity

Individuals with rare diseases encounter medical costs three to five times higher compared to those conditions, underscoring the financial burden associated with managing these often complex and chronic health issues.⁶ The economic impact of a delayed diagnosis can reach up to \$517,000 in avoidable costs per patient.⁷



Mental health challenges due to ongoing disease

Caregivers and patients, especially those in underrepresented communities, encounter substantial mental health challenges, including chronic stress, caregiver burnout, and heightened risk of anxiety and depression.⁸ The compounded effect of caregiving stress, financial barriers, and systemic discrimination results in a higher prevalence of mental health challenges, particularly in communities already facing structural barriers to healthcare.⁹

Marginalized groups are disproportionately affected by health disparities, resulting in longer diagnostic delays and poorer health outcomes.¹⁰ Investigating the extent of these disparities— particularly within specific subpopulations—and addressing health equity in rare disease care (including ultra-rare conditions like PNH) is crucial for developing targeted interventions and improving outcomes for rare disease patients.



⁵ <https://www.mdpi.com/1660-4601/20/6/4732>, https://rarediseases.org/wp-content/uploads/2020/11/NRD-2088-Barriers-30-Yr-Survey-Report_FNL-2.pdf

⁶ https://ncats.nih.gov/sites/default/files/NCATS_RareDiseasesFactSheet.pdf

⁷ <https://globalgenes.org/rare-disease-facts/>

⁸ <https://www.rarediseasediversity.org/research-resources>

⁹ *Id.*

¹⁰ https://rarediseases.org/wp-content/uploads/2020/11/NRD-2088-Barriers-30-Yr-Survey-Report_FNL-2.pdf

Understanding Paroxysmal Nocturnal Hemoglobinuria (PNH)



PNH is characterized by the destruction of red blood cells due to a deficiency of protective proteins, resulting in various severe symptoms and complications.¹¹ PNH is associated with other serious conditions such as aplastic anemia and myelodysplastic syndrome; leukemia; and Budd-Chiari syndrome, a rare liver disorder

caused by blood clots; this emphasizes the need for ongoing monitoring and management.¹² PNH weakens the immune system, making patients more susceptible to severe infections that can become life-threatening.¹³ PNH predominantly affects adults in their 30s and 40s, although it can occur at any age and across all races and genders.¹⁴ Approximately 400 to 500 new cases are diagnosed annually in the US.¹⁵

PNH presents several significant challenges for patients, primarily due to hemolysis and the destruction of red blood cells. Severe hemolysis can lead to fatigue; dysphagia; painful abdominal and esophageal spasms; and, in men, erectile dysfunction.¹⁶ Chronic hemolysis also increases the risk of blood clots, particularly venous thrombosis, which can result in life-threatening complications affecting organs like the liver, stomach, brain, and lungs.¹⁷ Additionally, all PNH patients experience bone marrow dysfunction, leukopenia, and thrombocytopenia.¹⁸ These complications make PNH a complex and life-threatening disorder that requires careful management.

PNH presents several significant challenges for patients, primarily due to hemolysis and the destruction of red blood cells.

¹¹ <https://rarediseases.org/rare-diseases/paroxysmal-nocturnal-hemoglobinuria/>

¹² [/d.](#)

¹³ <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC10653694/>

¹⁴ <https://rarediseases.org/rare-diseases/paroxysmal-nocturnal-hemoglobinuria/>

¹⁵ [/d.](#)

¹⁶ [/d.](#)

¹⁷ [/d.](#)

¹⁸ [/d.](#)

Getting the Right Diagnosis and Treatment

The diagnosis of PNH typically involves a combination of clinical evaluation; blood tests; and flow cytometry, a specialized technique used to detect the absence of protective proteins on the surface of red blood cells.¹⁹ Due to the rarity of the disease and its varied symptoms, PNH is often misdiagnosed or diagnosed late, leading to delays in treatment and a higher risk of complications.

Advancements in PNH research have led to FDA-approved treatments that significantly improve patient outcomes, although treatment is not curative. Treatment focuses on managing symptoms; preventing complications; and improving the patient's quality of life, extending their quality of life. Prior to the development of treatments for PNH, patients had a median survival range of 10 to 22 years.^{20,21}

Despite advancements in treatment, access to these advanced therapies and specialized care is limited in the United States. Many health insurers do not cover the high treatment costs incurred when managing PNH. Medicaid, Medicare, and private insurance providers often impose delays for approval of medications, sometimes due to limited coverage for orphan drugs.

The logo for the American Joint Committee on Cancer (AJMC) is displayed in a serif font within a purple rectangular box.

Many insurance companies prioritize reimbursement for less expensive medications. Drugs with multiple indications often are not covered, making it more difficult for patients living with medically complex conditions to find treatment.

People living with PNH may need other costly treatments such as blood transfusions, anticoagulant therapy, and bone marrow transplants for severe cases.²³ These treatments are critical in managing PNH because they address both the underlying cause and the severe symptoms associated with the disorder, offering patients the potential for a more stable and manageable condition. Yet for many PNH patients, including those who are



¹⁹ [id.](#)

²⁰ <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC10032435/>

²¹ <https://rarediseases.org/rare-diseases/paroxysmal-nocturnal-hemoglobinuria/>

²² <https://www.ajmc.com/view/predictors-of-orphan-drug-coverage-restrictions-in-medicare-part-d>

²³ <https://rarediseases.org/rare-diseases/paroxysmal-nocturnal-hemoglobinuria/>

uninsured or underinsured, financial barriers may prevent them from getting access to the help that they need.

Supporting Individuals with PNH

Support for an individual living with PNH may come from a variety of sources.

Patient navigators within health systems:

1

Originally developed for cancer care and adapted for other chronic conditions, patient navigators enhance healthcare access, facilitate patient-provider communication, and provide psychosocial and financial support. While often knowledgeable about navigating cancer in general, navigators may lack the specific knowledge required to adequately address the unique challenges faced by rare disease patients, including those living with PNH.

Family and friend caregivers:

2

Caregivers play a vital role in providing emotional and practical support, often coordinating healthcare appointments and managing medications. Access to specialized healthcare teams and advocacy organizations can further empower patients and families, ensuring they receive comprehensive care and resources tailored to their unique needs.

Patient Advocacy Organizations:

3

Patient advocacy organizations and assistance programs are pivotal in providing education, advocacy, and financial assistance to help patients navigate the complexities of PNH care. These organizations can help raise awareness, provide patient resources, and promote clinical research.

- [Aplastic Anemia and MDS International Foundation \(AAMDSIF\)](#)
- [The National Organization for Rare Disorders \(NORD\)](#)
- [PNH Global Alliance](#)
- [PNH Support](#)

Advancing Research & Clinical Trials



The Global PNH Patient Registry is an international registry, sponsored by the Aplastic Anemia and MDS International Foundation, created by patients for patients that collects data directly from patients regarding their experience with PNH and its impact on their quality of life. This patient-reported information can then be used to enhance the understanding of PNH, identify gaps and improve the care of patients with PNH, and empower and unite the PNH community.

The International PNH Interest Group (IPIG) PNH Registry is an international observational database collecting health information about PNH patients over time from healthcare providers. The IPIG PNH Registry aims to collect data about all PNH patients including clinical outcomes, patient reported outcomes and health resource usage, as well as long term safety data for patients treated with anti-complement therapies.

The National PNH Service Registry in the UK is an international research database that collects anonymized data on patients with PNH worldwide. It is intended to gather information about the natural history and management of patients with PNH.

In the US, various research and clinical databases also contribute to understanding PNH through detailed patient and clinical data. Collectively, the registries play a pivotal role in enhancing our understanding of PNH and improving patient outcomes through comprehensive data collection and analysis. The resulting initiatives aim to improve comprehension of PNH and inform the development of equitable care strategies.

Several pivotal studies have significantly influenced the landscape of PNH treatment and understanding. The TRIUMPH Study and the SHEPHERD Study were instrumental in establishing the efficacy and safety of complement inhibitors for managing PNH, demonstrating substantial reductions in hemolysis and improvements in patients' quality of life. Moreover, the ALXN1210 Clinical Trials explored an advanced complement inhibitor, revealing promising results with a more convenient dosing schedule, thus enhancing treatment options. The REACH Study provided crucial insights into the substantial impact of PNH on patients' daily lives, underscoring the importance of effective symptom management. More recently, the APPLY-PNH and APPOINT-PNH trials have advanced research by evaluating novel therapies and their potential benefits for PNH patients. These trials imparted a broader understanding of PNH and its management, offering hope for improved treatment strategies and patient outcomes. These studies underscore the ongoing advancements in PNH research, aiming to enhance therapeutic approaches and address unmet needs in patient care.

Addressing Health Inequities for People with PNH

Disparities in the care and treatment of PNH are present across the disease journey. Studies highlight that individuals from disadvantaged and marginalized groups – such as those of lower socioeconomic backgrounds, non-English speakers, and racial and ethnic minorities – experience greater challenges in receiving timely and effective care. This gap is exacerbated by institutional biases, which limit access to care and undermine patient trust.

Without treatment, the 10-year mortality rate for PNH is approximately 29%.

Without treatment, the 10-year mortality rate for PNH is approximately 29%.²⁴ Symptoms and complications associated with PNH, such as thrombosis, are particularly concerning during pregnancy, in surgery, or in high-altitude environments, requiring specialized care and careful management. These life-threatening risks make early diagnosis and equitable access to care critical. Unfortunately, gaps in treatment availability remain.

Challenge 1: People with PNH may not receive timely or appropriate diagnosis, treatment, or care due to the ultra-rare nature of the disease.

Delays in diagnosis are a significant issue, leading to long therapy durations that particularly impact those who face barriers in access to timely healthcare.²⁵ PNH patients often experience severe complications such as chronic hemolysis, thrombosis, and significant fatigue. Thrombosis is a major cause of morbidity and mortality in PNH patients because it often leads to strokes, pulmonary embolism, and other life-threatening events. Chronic fatigue and weakness can limit daily functioning, causing difficulties with engaging in work and routine activities.

Moreover, there is a pressing need for more research to better understand these disparities and their impact on PNH patients. Increased research funding, diverse clinical

trials, and research participation that includes contributions of patient- and health provider-reported data to PNH registries are essential to provide a clearer picture of the populations most affected by these inequities. Identifying these gaps can guide future efforts in reducing disparities and improving outcomes for all PNH patients.

Complement inhibitors are critical for treating PNH because they help prevent the destruction of red blood cells central to the disparities and their impact on PNH patients. Increased research funding, diverse clinical trials, and research participation that includes contributions of patient- and health provider-reported data to PNH registries are essential to provide a clearer

²⁴ <https://www.jmcp.org/doi/10.18553/jmcp.2020.26.12-b.s3>

²⁵ <https://pubmed.ncbi.nlm.nih.gov/29721255/>

picture of the populations most affected by these inequities. Identifying these gaps can guide future efforts in reducing disparities and improving outcomes for all PNH patients.

Complement inhibitors are critical for treating PNH because they help prevent the destruction of red blood cells central to the disease's pathology.²⁶ However, access to such advanced therapies remains inequitable. Socioeconomic factors, geographical barriers, and gaps in healthcare coverage contribute to disparities in accessing these life-saving treatments. Accessing FDA-approved medications often requires specialist visits, which can be difficult to schedule due to a shortage of PNH experts and complex insurance processes.

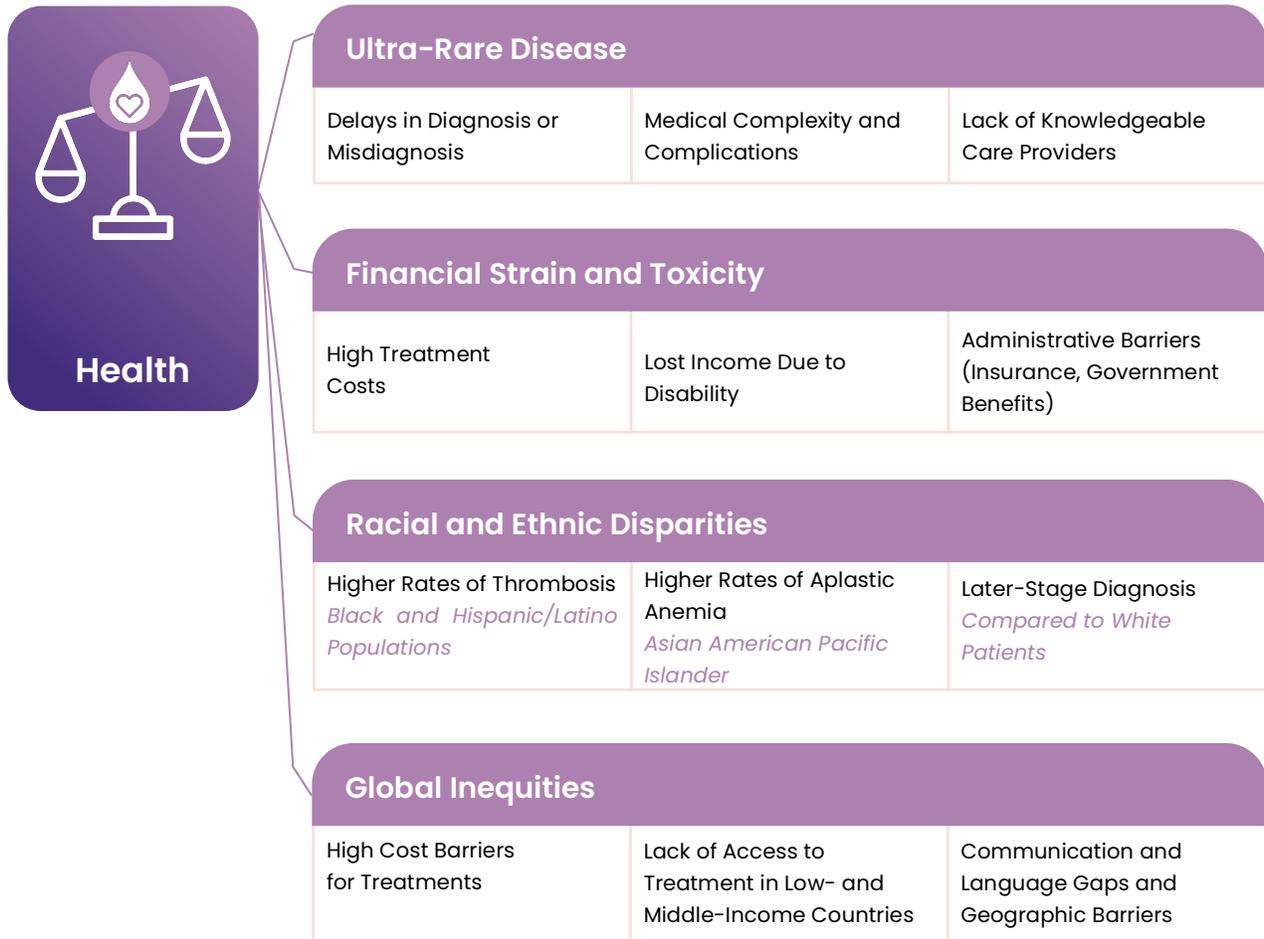
Additionally, the cost of treatments, including infusions and recurring visits, places a financial burden on patients, impacting their ability to adhere to prescribed therapies and thereby affecting treatment outcomes.

Without more equitable access, many PNH patients are left unable to benefit from the advancements in care that could drastically improve their prognosis.

Patients frequently encounter obstacles when seeking medication approvals and navigating limited drug supplies, which can disrupt their treatment routines. Additionally, patients often find themselves advocating for their needs and sharing new treatment information with healthcare providers, highlighting gaps in access and provider awareness. Effective communication between patients and providers is essential for managing PNH, yet communication gaps persist in medical teams, such as between hematologists and primary care physicians. Biases and a lack of shared decision-making often impede productive communication. Providers may not fully consider both medical and non-medical factors when making care decisions, thereby exacerbating disparities and impacting the quality of care.

²⁶ <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC10112829/>

Figure 1: Compounding Health Inequities Facing PNH Patients



The complexity of PNH, combined with the variability in individual responses to treatments, makes symptom management particularly challenging. Inadequate management of symptoms like fatigue can diminish quality of life and adversely affect overall health outcomes. Moreover, the absence of standardized tools for assessing the quality of life in PNH patients complicates efforts to identify and address specific issues impacting patient well-being. This lack of standardized assessment tools hinders the development of tailored care plans that could improve patient outcomes.

Challenge 2: Once diagnosed, financial strain may prevent people with PNH from getting access to the care they need.

Living with hematological conditions like PNH can result in severe functional limitations. Many PNH patients may need to access employment protections that allow them to manage their side effects at work through reasonable accommodations, or to take time off work and replace lost wages through state or federal disability insurance, such as Social Security Disability Income (SSDI) or Supplemental Security Income (SSI). To qualify, individuals must demonstrate significant impairments in bodily



function, such as recurrent episodes of hemolysis, severe anemia, or frequent hospitalizations due to infections.

In many cases, the criteria to qualify for benefits emphasizes that applicants must present detailed medical evidence, including laboratory results and clinical findings, to substantiate the extent of their disability. Disparities arise from the complexity of gathering sufficient medical documentation and accessing specialist care, which may disproportionately impact underserved populations.²⁷ This can create challenges in qualifying for benefits, especially when patients with rare conditions like PNH face

barriers in navigating the healthcare system and accessing comprehensive testing.

In addition to supplementing lost income because of the disease's impact, access to advanced therapies remains inequitable. Many patients cannot afford the high costs of treatment, which include infusions, routine visits to specialized providers, and the associated impacts on work and personal life. Medications, such as oral treatments, are significantly expensive, and the cost of over-the-counter (OTC) medications adds up due to the toll typical common illnesses take on PNH patients. Additionally, economic stability and education play a role in access to care. Patients may face difficulties in taking time off work for medical appointments and experience impacts on work and education continuity due to their condition.

In 2023, RDDC collaborated with NORD to conduct a groundbreaking national survey with over 2,800 participants to highlight barriers faced by underrepresented patients with rare diseases in accessing care. This research uncovered critical insights into healthcare access challenges, identifying several mental health issues. These included feelings of being overwhelmed, stressed, anxious, and hopeless, as well as financial strain preventing healthcare access (i.e. delayed and forgo) among patients and caregivers. These concerns are particularly severe among disadvantaged and marginalized groups, including LGBTQIA+ and lower socioeconomic individuals, with up to 72% reporting feeling overwhelmed.²⁸

²⁷ <https://www.ssa.gov/disability/professionals/bluebook/7.00-HematologicalDisorders-Adult.htm>

²⁸ <https://www.rarediseaseiversity.org/research-resources>

Challenge 3: Patients from diverse backgrounds face unique disparities in identifying and treating PNH.

Patients who are Black, Hispanic/Latino, and Asian American Pacific Islander face an increased risk of health disparities in the identification and treatment of PNH.



Black and Hispanic/Latino patients experience higher incidences of thrombosis compared to other racial groups, which is the leading cause of death for PNH patients.²⁹ Asian American Pacific Islander patients exhibit greater rates of aplastic anemia, often coexisting with PNH.

White patients in the US tend to be diagnosed younger and present with more classic PNH symptoms such as hemoglobinuria and infection. In contrast, patients who are Asian American Pacific Islander are often older at diagnosis and exhibit more bone marrow dysfunction.³⁰ This increased risk is attributed to genetic and environmental factors, emphasizing the need for targeted strategies to manage and prevent thrombotic complications in these patients. PNH patients using different treatments experience variations in healthcare resource utilization and

associated costs, which can exacerbate disparities in access to optimal treatment.³¹

Systematic issues, including policies influenced by social determinants of health (SDOH), contribute to disparities in PNH care. Access to healthcare for individuals with PNH is significantly impacted by SDOH, such as race, ethnicity, religion, sexual orientation, gender identity, age, disability, socioeconomic status, and geographical location. These determinants (or drivers) exert a substantial impact on a broad spectrum of health, functioning, and quality-of-life outcomes, playing a pivotal role in individuals' capacity to access high-quality healthcare.³²

For example, variability in Medicaid coverage and expansion across states impacts the availability of healthcare services for PNH patients, with state politics influencing the scope of coverage and access. Patients in rural areas face

²⁹ <https://www.thieme-connect.de/products/ejournals/abstract/10.1160/TH04-06-0391>; <https://emedicine.medscape.com/article/207468-overview?form=fpf>

³⁰ <https://emedicine.medscape.com/article/207468-overview?form=fpf>

³¹ <https://pubmed.ncbi.nlm.nih.gov/36633725/>

³² [https://health.gov/healthypeople/priority-areas/social-determinants-health#:~:text=Social%20determinants%20of%20health%20\(SDOH,of%20life%20outcomes%20and%20risks.](https://health.gov/healthypeople/priority-areas/social-determinants-health#:~:text=Social%20determinants%20of%20health%20(SDOH,of%20life%20outcomes%20and%20risks.)

substantial challenges due to long travel distances to specialized care centers and the limited availability of telehealth options. This geographical disparity exacerbates difficulties in accessing timely and appropriate care.

The lack of standardized clinical guidelines for PNH results in inconsistent care and outcomes across healthcare settings. The underrepresentation of diverse populations in clinical trials and research limits insights into inequities and hampers the development of effective treatments and care strategies.

Socioeconomic and cultural factors further complicate PNH care. Cultural barriers often arise from differing beliefs, values, and practices related to health, leading to misunderstandings between healthcare professionals and patients. When healthcare providers do not recognize or respect their patients' cultural backgrounds, it can create an environment of alienation that makes patients less likely to seek care or adhere to treatment recommendations. Health literacy, numeracy challenges, and

language barriers also impede effective communication between patients and healthcare providers, leading to misunderstandings that affect treatment fidelity and outcomes.³³

Community resources and support systems shaped by SDOH are critical in impacting patient care and outcomes. Some immigrants face additional challenges such

Strong support networks, including mental health support, emotional assistance, and advocacy, are essential for managing PNH effectively.

as language barriers, cultural differences, legal restrictions, and financial obstacles, further impacting their access to care and treatment. Addressing these barriers is crucial for improving health equity in PNH care and ensuring that all patients receive comprehensive, effective treatment.

Challenge 4: Global inequities make it difficult for people with PNH to receive care and treatment equitably across nations

Global access to PNH treatments varies significantly. PNH treatments are widely available in many countries, though their high cost can limit access. Newer treatments, including those inhibiting different immune system proteins, offer additional options but are also subject to regional

³³ <https://health.gov/healthypeople/priority-areas/social-determinants-health/literature-summaries/language-and-literacy>

availability. Supportive treatments such as anticoagulants, blood transfusions, and iron supplements are more universally accessible.³⁴

Advanced therapies are often unavailable in low- and middle-income countries, parts of sub-Saharan Africa, and certain regions in Asia and Latin America, reflecting disparities in healthcare resources and economic constraints.

Some key challenges contributing to disparities in PNH care are the limited availability of healthcare professionals with PNH expertise, lack of specialists or specialty clinics in rural areas and developing countries, communication gaps between medical teams, and geographical barriers.

³⁴ <https://pnhuk.org/what-is-pnh/current-treatments/>; <https://pnhglobalalliance.org/dealing-with-pnh/>

Recommendations for Enhancing Health Equity in PNH Care

Given these considerations, the RDDC and Links2Equity convened the PNH Health Equity Advisory Committee. After reviewing the research, the Committee offered the following recommendations to improve access to equitable, timely, and appropriate care for people living with PNH. A full list of these recommendations is included in Appendix A.

1. Improve Equitable Access to Treatment

Streamline treatment approval processes by simplifying prior authorizations and reducing non-medical switching, focusing on underinsured and low-income patients with healthcare providers and insurers taking the lead in implementation, and with patient advocacy organizations and policymakers supporting these efforts through advocacy for policy change (*Appendix A: Recommendation 1.4*).

Expand access to care by prioritizing telehealth and mobile clinics to serve rural and marginalized communities with healthcare systems and providers taking the lead in the implementation of these efforts. Community-based organizations and internet service providers should support these efforts by facilitating access to low-cost or free internet services for populations lacking access. (*Appendix A: Recommendation 6.1, 6.2*).

2. Enhance the Patient and Caregiver Experience

Integrate shared decision-making aids by developing multilingual and accessible decision-making tools for patients and caregivers with healthcare providers and patient advocacy organizations leading the development and integration of decision-making aids (*Appendix A: Recommendation 4.1*).

Establish a PNH coalition to advocate and support patients and caregivers, with a focus on underserved groups (*Appendix Recommendation 4.2*). Patients, caregivers, and patient advocacy organizations should lead the formation of a PNH coalition to advocate for patient and caregiver needs. Additionally, policymakers can support such a coalition by offering resources, expertise, and legislative backing.

3. Reduce Financial Barriers

Expand financial assistance programs to increase financial support for low-income and underinsured PNH patients, targeting costs related to treatment, travel, and other expenses (*Appendix A: Recommendation 2.1, 2.1.2*). Pharmaceutical and philanthropic companies can lead in expanding financial assistance programs for PNH treatments, particularly for low-income and uninsured populations. Healthcare providers and patient advocacy groups can provide support by connecting patients with available resources and raising awareness.

Advocate for comprehensive insurance coverage by working with insurers to reduce out-of-pocket costs for PNH treatments. Insurers and policy advocacy groups should lead efforts to reduce out-of-pocket expenses and improve insurance coverage for PNH treatments (*Appendix A: Recommendation 2.2*). Healthcare providers and patient coalitions can support these efforts by providing data on financial burdens and advocating for reforms.

4. Enhance Provider Education

Implement a comprehensive continuing medical education (CME) training for providers by focusing on cultural competence, gender sensitivity, pain management, mental health integration, and improving communication between healthcare teams and patients (*Appendix A: Recommendation 3.1, 5.2*). Medical associations and training institutions should lead in developing cultural competence and gender sensitivity training for healthcare providers. Patients, caregivers, and patient advocacy organizations can support training development by providing input on patient experiences and diverse perspectives.

Provide ongoing education on PNH treatments to medical teams, ensuring providers are kept up to date on the latest treatment options and clinical guidelines for PNH (*Appendix A: Recommendation 3.1*). Healthcare systems and medical institutions should lead efforts to ensure ongoing education for medical teams on the latest PNH treatments and clinical guidelines. Patient advocacy organizations, medical associations, and pharmaceutical companies can support this by funding educational initiatives and providing updated treatment information.

5. Promote Equity in Research

Increase diversity in clinical trials by advocating for inclusive trial designs that reflect diverse racial, gender, and socioeconomic groups (*Appendix A: Recommendation 7.1*). Pharmaceutical companies and research institutions should lead in designing inclusive clinical trials that address the needs of diverse populations. Community-based organizations and patient advocacy groups can support this by providing clinical trial education and outreach assistance in recruitment efforts and building trust among marginalized populations.

Engage community organizations in research by collaborating with community organizations to build trust and encourage trial participation among underserved populations (*Appendix A: Recommendation 7.2*). Community organizations and research institutions should lead the collaboration efforts to engage underserved populations in research. Healthcare providers and patient advocacy organizations can support research equity by encouraging patient participation and providing culturally sensitive outreach.

The implementation of these recommendations is vital in enhancing the overall care, support, and outcomes for individuals affected by PNH. By improving accessibility to FDA-approved treatments, addressing financial barriers, enhancing provider awareness and education, and patient-centered approach to managing this condition.

It is essential for stakeholders, including healthcare providers, pharmaceutical companies, insurers, patient advocacy organizations, and policymakers, to collaborate and take proactive steps in realizing these recommendations, ultimately making a meaningful difference in the lives of PNH patients and their families.



Strategies for Advancing Health Equity in PNH

The following actionable strategies are offered to achieve the PNH Health Equity Recommendations. A multi-stakeholder approach is crucial to improving access to FDA-approved PNH treatments and addressing broader health equity barriers.

Strategy 1: Empower Shared-Decision Making



Patients and caregivers should actively participate in shared decision-making by using tools that facilitate discussions on treatment options and non-medical factors such as emotional well-being. This collaborative approach mirrors successful efforts in diseases like multiple sclerosis, where patients' lived experiences have informed tailored treatment plans.³⁵

Advocacy, in which patients and caregivers engage in policy efforts to improve insurance coverage, address pain management access, and remove barriers to treatment, is also essential. This is indicative of past HIV/AIDS advocacy efforts that have secured equitable treatment access.³⁶

Strategy 2: Educate Healthcare Providers on PNH Treatment and Care



Healthcare providers need regular training on the latest PNH treatments, clinical guidelines, the integration of mental health, and comprehensive pain management, paired with cultural competence training to mitigate biases in care. Models such as the Hepatitis C Provider Education Program, which included patient-centered care and cultural awareness, offer a template for training that enhances provider-patient interactions.³⁷

Providers can also establish collaborative partnerships with patients to ensure treatment strategies incorporate patient voices; this has been effective in breast cancer care through initiatives like the patient-provider collaborative learning forums. Collaborative models that integrate patient perspectives into care, such as the Patient-Centered Outcomes Research Institute (PCORI), have proven successful as well.³⁸ Integrating mental health into PNH care is another key area. Having a holistic model, like the integration of mental health services in care,

³⁵ [https://www.msard-journal.com/article/S2211-0348\(23\)00593-X/fulltext](https://www.msard-journal.com/article/S2211-0348(23)00593-X/fulltext)

³⁶ <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4605825/>

³⁷ <https://pubmed.ncbi.nlm.nih.gov/17542458/>

³⁸ <https://www.pcori.org>

can help providers address the emotional and psychological challenges associated with chronic illness.³⁹

Oncology care has been inclusive of nurses, social workers, or patient advocates who are trained to assist patients in navigating complex healthcare systems, addressing disparities in treatment access, and providing personalized support throughout their care journey. For instance, the Academy of Oncology Nurse & Patient Navigators (AONN+) has developed extensive training and certification programs for patient navigators, improving patient outcomes through tailored support.⁴⁰ The development of PNH-specific training programs can support the inclusion and integration of PNH patient navigators.

Strategy 3: Strengthen and Support a Multi-Stakeholder PNH Coalition



Rare patient advocacy organizations strengthen health equity efforts in care by building disease-specific coalitions dedicated to health equity and underserved groups. An example from the cystic fibrosis community shows how patient advocacy organizations effectively improve patient care and streamline research participation by engaging patients and caregivers in every aspect of the process.⁴¹ These organizations and coalitions can also work with policymakers to advocate for Medicaid expansion and other legislative reforms; this is much like the advocacy for expanded coverage for ALS patients, which has successfully reduced out-of-pocket costs for treatments.⁴² Community education efforts focused on insurance navigation and financial support are essential, as shown in the success of initiatives for hemophilia, where patient groups raised awareness of and improved access to financial aid for treatment.⁴³

Strategy 4: Prioritize Inclusive Clinical Trial Designs and Representative Research



Researchers must prioritize inclusive trial designs that reflect the diverse racial, gender, and socioeconomic backgrounds of PNH patients. HIV/AIDS research has successfully implemented such inclusive strategies, ensuring trials meet the needs of varied patient groups and generating more equitable treatment outcomes.⁴⁴

³⁹https://professional.diabetes.org/sites/default/files/media/psychosocial_care_for_people_with_diabetes_dempaire.pdf

⁴⁰<https://aonnonline.org>

⁴¹<https://www.cff.org/get-involved/how-we-advocate-cf-community#:~:text=We%20help%20ensure%20states%20allocate,high%2Dquality%2C%20specialized%20care.>

⁴²<https://www.als.org/blog/advocating-break-down-health-insurance-barriers>

⁴³<https://www.hemophiliafed.org/resource/non-hfa-financial-assistance/>

⁴⁴<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC8650944/>

Collaboration with community-based organizations, as done in SCD research, will ensure recruitment efforts are culturally sensitive and tailored to engage underserved populations.⁴⁵

The American Society for Hematology (ASH) is continuing its efforts to achieve greater equity through its initiative, *A Path of Progress*.⁴⁶ The initiative discusses various strategies, including targeted educational opportunities and support programs aimed at underrepresented groups. By fostering a more inclusive environment, ASH aims to address disparities and ensure a diverse workforce that reflects the broader patient population. These efforts are part of the organization's broader mission to advance health equity and improve outcomes in hematology.

Strategy 5: Reduce Financial Barriers to PNH Care



Legislators and policymakers must support state-level Medicaid expansion and advocate for policies that streamline insurance processes, such as prior authorization reforms and protections for underinsured populations. Similar advocacy in the treatment of rare diseases like Gaucher disease has reduced bureaucratic barriers and improved patient access to necessary treatments.⁴⁷ Additionally, implementing cultural competency training and unbiased pain management protocols for healthcare providers, as seen in the opioid epidemic response, will help address disparities in care delivery.⁴⁸

Insurers also play a critical role by offering comprehensive coverage for PNH treatments and reducing the administrative burdens patients face when seeking approval for care. Programs for hemophilia have set an example by working closely with patient advocacy groups to create more flexible and accessible insurance programs.⁴⁹ Expanding these efforts, especially for individuals living with lower socioeconomic status who face marginalization, will further promote equity in rare disease care.

Lastly, the North Carolina Sickle Cell Syndrome Program exemplifies effective state-level management of a rare disease. It offers comprehensive services including genetic counseling, newborn screening follow-up, care coordination, referrals, and educational materials. Through a systematic statewide approach, the program utilizes Regional Sickle Cell Educator Counselors, comprehensive federally qualified community health centers, medical centers, and community-based organizations to provide accessible care. The reimbursement program assists with medical services for eligible patients through the purchase of medical care services, covering outpatient and emergency visits, hospitalization, and more, with prior authorization required. The

⁴⁵ <https://www.ncbi.nlm.nih.gov/books/NBK566476/>

⁴⁶ <https://ashpublications.org/ashclinicalnews/news/7894/A-Path-of-Progress-ASH-Works-to-Increase-Diversity>

⁴⁷ <https://www.gaucherdisease.org/blog/category/insurance/>

⁴⁸ <https://www.cdc.gov/overdose-prevention/hcp/clinical-guidance/index.html>

⁴⁹ <https://www.bleeding.org/advocacy/state-priorities/medicaid-state>

plan helps mitigate care disruptions and improves medication access.⁵⁰ There is long-standing collaboration with the Governor's Council on Sickle Cell Disease that ensures the program addresses evolving needs. This integrated model improves outcomes and supports families, embodying a commitment to quality care, community engagement, and health equity.⁵¹

Noteworthy examples from other disease states provide valuable insights into addressing health equity in PNH. By drawing from the successes of other diseases and involving stakeholders across the healthcare spectrum, we can significantly improve access to treatments and quality of life for PNH patients.

Conclusion

PNH is a challenging, life-altering condition that requires careful attention to the people and families most impacted. A healthcare landscape that ensures equitable access to care for all individuals living with PNH, regardless of their background or circumstances, is possible.



Given the challenges facing the PNH community, the RDDC, Links2Equity, and the PNH Health Equity Steering Committee recognize the urgent need for action to address the significant disparities in PNH care.

⁵⁰ <https://ncsicklecellprogram.dph.ncdhhs.gov/services.asp> ; <https://ncsicklecellprogram.dph.ncdhhs.gov/Documents/SC-FormularyList.pdf>

⁵¹ <https://ncsicklecellprogram.dph.ncdhhs.gov/#:~:text=Through%20the%20implementation%20of%20a,sickle%20cell%20educational%20materials%20and>



Appendix A: Comprehensive Recommendations for Enhancing Health Equity in PNH Care

1. Recommendation 1: Improve Accessibility to FDA-Approved Treatments for PNH

- 1.1 Provide routine training for healthcare providers on the latest PNH treatments, clinical guidelines, and pain management strategies. These trainings should be facilitated by PNH specialists and should include cultural competence training to address biases and enhance communication.
- 1.2 Expand provider access to pain specialists to address limitations in comprehensive pain treatment knowledge within medical teams.
- 1.3 Implement advocacy efforts that address the removal of barriers for PNH patients to certain treatment options that may be linked to system policies or concerns about addiction.
- 1.4 Implement advocacy efforts that support streamlining approval processes by simplifying prior authorizations and reducing non-medical switching, and for policies that protect lower socioeconomic and underinsured populations.
- 1.5 Implement community education initiatives to address patient knowledge regarding insurance options and navigation, alongside advocating for the removal or reduction of co-pay accumulators and co-pay maximizers to enhance access to affordable care.

2. Recommendation 2: Address Financial Barriers

- 2.1 Expand financial assistance program support for PNH treatment costs, including infusions, travel, and recurring visits. Partner with pharmaceutical companies and nonprofits to secure grants or subsidies.
 - 2.1.1. Encourage pharmaceutical and biotech companies to establish PNH patient focus groups to gain insights into the support needed for reimbursement through Hub Programs and to educate patients on the available support.
 - 2.1.2. Direct funding toward lower socioeconomic, underinsured, and uninsured populations, ensuring support for those disproportionately affected by high treatment costs. Increase education and awareness within these communities.
 - 2.1.3. Improve access to drug discount programs and specialty drugs and restrict co-pay accumulators and maximizers from taking discounts from vulnerable patients.
- 2.2 Advocate for comprehensive insurance coverage through strategic engagement with insurers to ensure comprehensive coverage for PNH treatments and advocate for legislative changes to reduce out-of-pocket costs.

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- 2.2.1. Patients and patient advocacy groups (PAG) can engage with insurers by advocating for policy changes, providing education, presenting data, forming partnerships, navigating claims, and building support networks to improve coverage for rare diseases.
 - 2.2.2. Support Medicaid expansion and other public insurance programs, particularly in states with large, marginalized populations.

3. Recommendation 3: Improve Provider Awareness and Education

- 3.1** Implement comprehensive training sessions for healthcare providers on the latest PNH treatments, clinical guidelines, and patient management strategies that are inclusive of cultural competence training to effectively address biases and enhance communication.
 - 3.1.1. Establish routine and robust continuing medical education (CME) training for healthcare providers on the latest PNH treatments, clinical guidelines, and gender sensitivity training to mitigate biases and ensure the patient voice is prioritized in management strategies. This education should be delivered by an unbiased third party such as PNH Specialists, rather than by industry representatives. Additionally, create provider and healthcare team peer-to-peer learning initiatives to foster knowledge-sharing and improve PNH care practices.
- 3.2** Establish patient-provider collaborative learning, and encourage and foster collaboration where providers actively integrate patient-shared knowledge about new treatments. Establish forums for shared learning, providing translation services for non-English speaking patients.
- 3.3** Establish learning initiatives engaging healthcare teams to enhance knowledge of PNH care. Form multidisciplinary care teams that include hematologists, mental health professionals, nutritionists, and social workers, enabling a holistic approach to address the complex needs of PNH patients.
- 3.4** Establish the integration of PNH care and mental health services to enhance provider knowledge and access to mental health clinicians in the care of PNH patients. Integration of services for PNH patients is paramount.

4. Recommendation 4: Enhance the Patient Experience and Empowerment

- 4.1.** Facilitate shared decision-making through the development of decision aids tailored to PNH patients to discuss medical and non-medical factors. Ensure tools are accessible in multiple languages and formats. Examples include a symptoms checklist, treatment options, and emotional support assessment/inventory.
 - 4.1.1. Implement continuous feedback loops as part of the shared decision-making process. This involves regular evaluations of patient progress, treatment effectiveness, and patient satisfaction with care. Use periodic assessments to



adjust care plans based on patient-reported outcomes and clinician insights. Feedback tools should be incorporated into decision aids, allowing for real-time updates and changes, ensuring decisions remain relevant and aligned with the patient's evolving needs. This approach will promote ongoing dialogue between patients and providers, ensuring personalized care that adapts over time.

4.2. Strengthen patient advocacy and support through the establishment of a PNH Coalition or PNH patient advocacy organization, in collaboration with PAGs, dedicated solely to improving PNH care:

4.2.1. Provide education and empowerment for caregivers, encourage patient advocacy organizations to create platforms for patients to share experiences and advocate for themselves, and ensure alignment among various PNH-serving organizations.

4.2.2. Strategically focus health equity efforts and outreach to underserved or underrepresented groups, providing culturally sensitive support.

4.2.3. Implement, fund, and collaborate on PNH research activities such as supporting patient recruitment, informing research priorities, and encouraging PNH registry participation.

4.2.4. Engage patients and caregivers in research participation and address the challenges of low numbers and limited knowledge of diversity in hematology clinical trials for PNH through peer-to-peer initiatives such as research ambassadors' programs.

4.3. Utilize social media as a strategic platform to raise awareness about PNH by creating targeted campaigns that encourage individuals to seek care, share valuable information about treatment accessibility, and amplify patient stories. Collaborate with social media influencers who have a strong connection to health advocacy to help reduce stigma associated with PNH, promote understanding of the condition, and foster a supportive community for patients and caregivers.

4.4. Develop training to support the inclusion of specialty trained patient navigators in PNH care. The inclusion of PNH patient navigators is essential for addressing barriers faced by patients with rare blood disorders like PNH. Focus patient navigation efforts on PNH patients from populations at increased risk for health inequities.

5. Recommendation 5: Reduce Communication Barriers, Address Bias and Stereotypes

5.1. Empower and educate PNH patients and caregivers to increase health literacy, numeracy, and language accessibility.

5.1.1. Deliver educational resources in multiple languages, and establish interpreter services.



5.1.2. Collaborate with community organizations to distribute materials and conduct workshops for patients and caregivers, emphasizing a comprehensive understanding of PNH, its symptoms, treatment options, and effective communication with support persons or caregivers.



- 5.2. Cultivate cultural competency through the provision of impactful and comprehensive cultural competency training for healthcare providers, medical teams, and all staff who interact with patients. Highlight the profound impact of race, gender, geography, and socioeconomic factors on care and address biases and stereotypes affecting care quality.
- 5.3. Address and mitigate prejudices in the medical system through the implementation of compelling medical system training opportunities on unconscious bias, with a specific focus on how it impacts patient interactions and outcomes. Collaborate with patients and caregivers to develop engaging programs with supplemental learning materials such as case studies and role-playing tailored to PNH care.
- 5.4. Advocate for the standardization of the diagnostic protocols to minimize the influence of race and gender on diagnosis and treatment. Ensure thorough evaluations for all patients.
- 5.5. Combat racial and gender disparities in care by advocating for the development of PNH-specific tools to assess pain and conduct regular evaluations of treatment outcomes by race and gender. Develop strategies to ensure equitable treatment and care for all patients.
- 5.6. Enhance pain assessment knowledge for providers through the development of culturally sensitive pain assessment and management training programs, ensuring that all patients' pain reports are acknowledged and treated appropriately.
- 5.7. Implement bias-free pain management protocols by establishing robust protocols to prevent racial and ethnic biases in pain management and consistently review practices to ensure equity.
- 5.8. Educate providers on gender bias through the development and facilitation of training to address gender bias and medical sexism, and ensure that female patients' concerns



are earnestly addressed. Include powerful discussions on intersectionality in the training and patient stories that highlight their experiences in care.

- 5.9. Promote gender-inclusive care by encouraging further development of gender-inclusive care practices and champion the development of guidelines addressing gender differences in PNH symptoms and treatments. Advocate fervently for research with diverse gender representation.

6. Recommendation 6: Overcome Geographical and Logistical Barriers

- 6.1. Prioritize the expansion of Medicare telehealth flexibilities in the Coronavirus Aid, Relief, and Economic Security Act (CARES) for PNH patients, particularly in remote areas, and ensure comprehensive insurance coverage. Additionally, providing low-cost or free internet access to marginalized populations is essential for equitable access to care.
- 6.2. Improve rural access to care by supporting the establishment of mobile clinics and satellite PNH and pain specialist offices, staffed by culturally competent providers, which is crucial to ensuring better access to care for PNH patients in rural areas.

7. Recommendation 7: Promote Equity in PNH Clinical Research

- 7.1. Increase diversity in research and clinical trials through advocacy for diverse populations in PNH clinical trials and ensuring that trial designs address the needs of multiple racial, gender, and socioeconomic groups. These are fundamental steps in promoting equity in clinical research.
 - 7.1.1. Patient advocacy organizations should seek funding from industry partners and grants to develop a clinical trial tool specific for PNH patients and share funds with community partners to expand access to clinical trial information with community providers and organizations.
- 7.2. Engage communities and community-based organizations as partners in trial design and recruitment, and to raise awareness about PNH and encourage research participation. Tailoring engagement efforts to be culturally sensitive is critical for building trust and participation.

8. Recommendation 8: Address Broader Socioeconomic and Policy Barriers

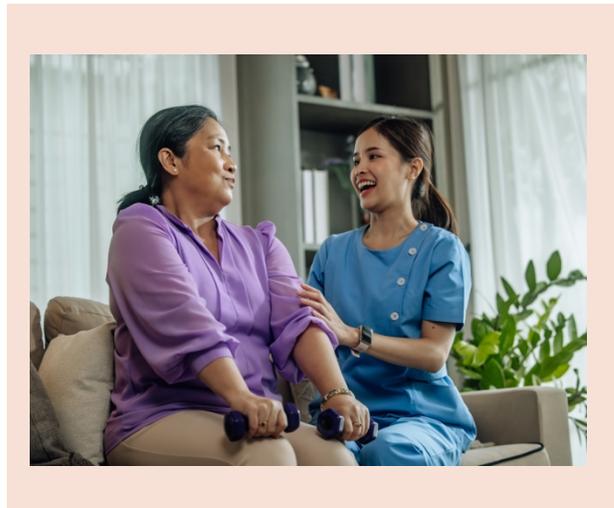
- 8.1. Advocate for Medicaid coverage & expansion for PNH treatment per state, through collaboration with policymakers with a focus on states with significant marginalized populations, which is crucial in addressing broader socioeconomic and policy barriers.
 - 8.1.1. Partner with local healthcare advocates and organizations to identify gaps in Medicaid coverage specific to PNH treatment to develop increased awareness among policymakers about the importance of including PNH-related services.

8.1.2. Implement outreach programs aimed at educating communities about the benefits of Medicaid expansion, particularly for rare diseases like PNH. Focus on sharing success stories from states that have expanded Medicaid and that have highlighted improved access to healthcare for marginalized populations to build public support and encourage policymakers to consider expansion as a viable option to address socioeconomic barriers.

8.2. Support PNH patient and caregiver knowledge and understanding of workplace and educational accommodations by providing outreach, education, and awareness of workplace and educational accommodations. Providing accommodation education for low-wage workers and students from marginalized backgrounds will help ensure equitable access to care and support.

9. Recommendation 9: Strengthen Community and Support Systems

9.1. Prioritize strengthening patient and caregiver support through the establishment of a robust patient advocacy network to support PNH patients and advocate for specific legislation. Encourage the development of patient support groups, both online and in person, involving caregivers and family members. Consider integrating mental health clinicians with lived experiences or whoserve the PNH and rare disease community.



9.2. Increase mental health support to PNH patients and caregivers through the development and implementation of peer support programs.

9.3. Integrate mental health services into PNH care and ensure accessibility to marginalized populations by offering services in multiple languages.

10. Recommendation 10: Address Immigrant-Specific Barriers

10.1. Support PNH patients and caregivers in navigating legal and financial challenges through needed supports that provide educational resources and prioritize support for immigrant PNH patients, including legal assistance and information on insurance options.

10.2. Advocate for culturally sensitive care that ensures healthcare providers are trained to understand and respect cultural differences. Advocate for partnerships with patient organizations and patient groups to assist in the development of these trainings with an emphasis on culturally appropriate educational materials for working with patients from immigrant communities.



11. Recommendation 11: Focus on Quality of Life and Symptom Management

- 11.1.** Prioritize the proactiveness of symptom management for PNH patients by encouraging providers to identify and address severe and persistent fatigue. Formulate care plans that encompass strategies to manage fatigue and enhance daily functioning.
- 11.2.** Enhance fatigue and pain assessment by incorporating social determinants of health into care plans, ensuring that patients from marginalized backgrounds receive comprehensive support that addresses their specific challenges. This includes access to transportation, secure housing, and adequate nutrition.