The Global PNH Patient Registry: Real-World Data on PNH Symptom Burden and Quality of Life

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INTRODUCTION

Current knowledge of the lived experience of individuals with Paroxysmal Nocturnal Hemoglobinuria (PNH) is informed by data collected during clinical trials. This information is therefore short-term and limited to individuals who choose to participate in an interventional trial. The Global PNH Patient Registry, sponsored by the Aplastic Anemia and MDS International Foundation, was designed to understand the impact of care and treatment effectiveness in real-world settings. It is the first patient-driven and patient-focused PNH registry with all data provided by individuals living with PNH or their caregivers.

AIM

We explored the longitudinal symptom burden and measures of quality of life (QoL) to quantify the impact of a diagnosis of PNH in this population living during this era of available complement inhibitor medications.

METHODS

Eligibility Criteria:
- Self-reported PNH diagnosis
- Ability to read English or Spanish
- Able to access the internet

The Global PNH Patient Registry is hosted on the National Organization for Rare Disorders' IAMRARE® platform – a web-based, secure portal for obtaining consent and entering patient-reported data. After initial survey completion, participants are notified every 6 months to update information.

Participants were asked to report their frequency of experience with 26 symptoms, rated on a 5-point Likert scale. In addition, PROMIS measures for seven QoL domains are included. Longitudinal data was examined by calendar year of submission for all participants and within participants among those with longitudinal data.

This analysis includes data provided between registry launch in April 2021 and December 31, 2024.

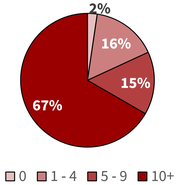
The study was reviewed and approved by North Star Review Board (IRB #NB100005).

RESULTS

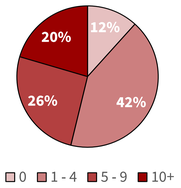
Study Population Characteristics	Number (%)
Age	
<18	3 (2%)
18 - <30	32 (18%)
30 - <50	77 (43%)
50 - <65	44 (25%)
65+	23 (13%)
Sex	
Male	61 (34%)
Female	117 (65%)
Race / Ethnicity (multi-select)	
American Indian / Alaskan Native	4 (2%)
Asian	26 (15%)
Black / African American	12 (7%)
Hispanic	20 (11%)
White	130 (74%)
Residence	
United States	122 (68%)
International	57 (32%)
Use Of Complement Inhibitor Medication	
Yes	90 (68%)
No	42 (32%)

List of Symptoms collected in the Registry	
Abdominal pain	Heart palpitations
Brain fog	Joint pain
Shortness of breath	Tingling fingers
Bruising easily	Weakness in arms and legs
Dark urine	Yellow skin and eyes
Fatigue	Blood clot
Headaches	Dysphagia
Lightheadedness	Erectile dysfunction
Muscle cramps in legs/feet	Esophageal spasms
Lower back pain	Nausea
Bone pain	Nerve pain/sensitive to touch
Gallbladder problems	Ringing in ears
Hair loss	Vertigo/dizziness

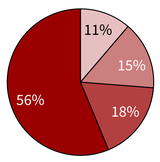
Number of Symptoms Study Participants Reported Experiencing at First Survey Submission



Percentage of Study Participants by Number of Symptoms they Report Experiencing Often or Always at First Survey Submission

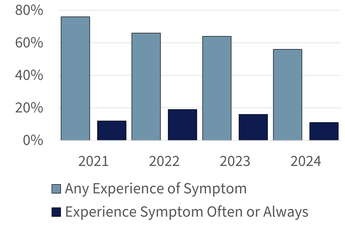


*Percentage of Study Participants by the Number of PROMIS Scores indicating Impairment of Quality of Life at First Survey Submission**

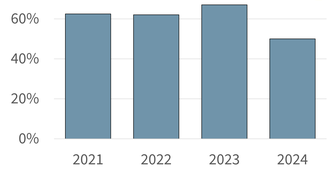


Key Findings:
- There is substantial symptom burden experienced by study participants, both for any experience and those experiencing symptoms often or always.
- Quality of life is impaired for most study participants.
- There is the suggestion of decreasing symptom burden over time but no indication of improvements in quality of life.
- Among participants with multiple symptoms and quality of life assessments, a small percentage report a change in symptom burden or quality of life impairment.

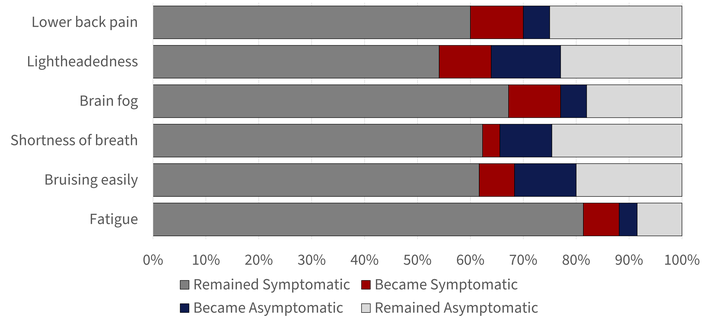
Percentage of Participants Reporting Experiencing Ten or More Symptoms by Callendar Year among All Survey Submissions



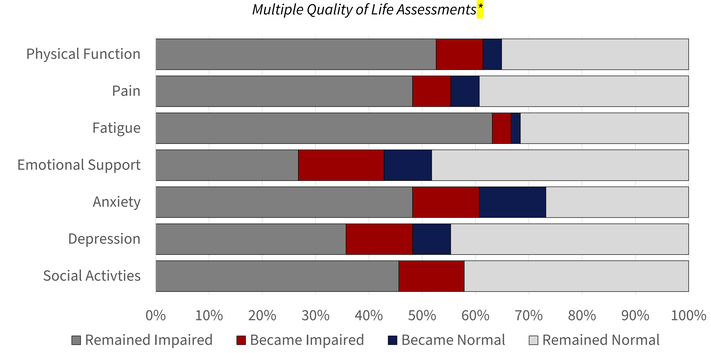
*Percentage of Participants Reporting Impairment in 3 or more PROMIS Scores by Callendar Year among All Survey Submissions**



Distribution of Reported Symptom Experience for the Six Most Commonly Reported Symptoms Among Individuals with Multiple Symptom Assessments



*Distribution of Reported Quality of Life Experience for Seven PROMIS Scores Among Individuals with Multiple Quality of Life Assessments**



*These graphs were updated since they were presented at the ASH 2025 conference to correct errors in the calculation of the PROMIS scores.

CONCLUSIONS

In this current population, with a substantial proportion reporting treatment with a complement inhibitor, a diagnosis of PNH was associated with substantial symptom burden and impairment of quality of life. This preliminary investigation of longitudinal trends suggests some potential decreases over time but, for most participants, substantial burden and impairment persists.

Next steps will be to assess the impact looking at more granular changes in symptoms and QoL and the impact of treatment and other demographic and disease factors to characterize the patterns over time. These data provide an important resource for understanding the real-world, lived experiences of individuals with PNH and to investigate long-term outcomes as care and treatment for this disease continues to evolve.

ACKNOWLEDGEMENTS

AAMDSIF extends our sincere appreciation to everyone who has made it possible to develop the Global PNH Patient Registry:
- ✓ The PNH Patient Community for participating in the Registry, completing surveys and sharing their experiences;
- ✓ The researchers and clinicians who are dedicating their time and expertise to people with PNH and for helping us raise awareness of this critical initiative;
- ✓ The Global PNH Patient Registry Patient Steering Committee and Medical Advisory Committee for their guidance and consultation;
- ✓ The industry sponsors whose support has enabled us to create and manage the Registry; and
- ✓ The AAMDSIF Board of Directors for their commitment to the mission and growth of the Registry.

For questions and to learn more about the PNH Registry, please contact us at: pnhregistry@aamds.org.