The Global Paroxysmal Nocturnal Hemoglobinuria (PNH) Patient Registry: Highlights of the Data from the First Three Years

D Araten[1], AG Kulasekararaj[2], J Nishimura[3], P De Latour[4], IC Weitz[5], A Candelaria[6], A Fink[7], A Houk[6], EW Kottke[7], and BA Wong[6]

[1]NYU Cancer Institute, New York, NY; [2]King's College Hospital-NHS Foundation Trust, NIHR / Wellcome King's Clinical Research Facility, and King's College London, London, United Kingdom; [3]Osaka University Graduate School of Medicine, Osaka, Japan; [4]BMT unit, Hôpital Saint-Louis, Paris, France; [5]Divisionof Hematology, Keck School of Medicine of USC, Los Angeles, CA; [6] Aplastic Anemia & MDS International Foundation, Rockville, MD; [7]National Organization for Rare Disorders, Quincy, MA

INTRODUCTION

Paroxysmal Nocturnal Hemoglobinuria (PNH) is a rare, acquired, hematologic disorder characterized by chronic complement-mediated hemolysis and associated with a mutation in the PIG-A gene, thrombosis, and immune-mediated marrow failure.

The Global PNH Patient Registry, initiated in 2021, is the first patient-driven and patient-focused PNH registry. The Aplastic Anemia and MDS International Foundation leads The Global PNH Patient Registry with input from patients and caregivers along with hematologists and researchers. All data are provided by individuals with PNH or their caregivers.

AIMS

The goals of the Global PNH Registry are to:

1. Enhance the understanding of PNH
2. Identify gaps and improve the care of patients with PNH
3. Empower and unite the PNH community.

METHODS

Inclusion Criteria

- ✓ Self reported diagnosis of PNH
- ✓ Able to read English
- ✓ Access to a computer or mobile device and the internet

Data Collection

Registry is hosted on the National Organization for Rare Disorder's IAMRARE® Platform

- ✓ Web-based
- ✓ Secure
- ✓ Obtain consent and enter patient-reported data

Data included in the registry includes:

- ✓ Demographics
- ✓ Diagnosis
- ✓ History of medications, procedures and supplements
- ✓ Current symptoms
- ✓ Health care utilization
- ✓ Quality of life

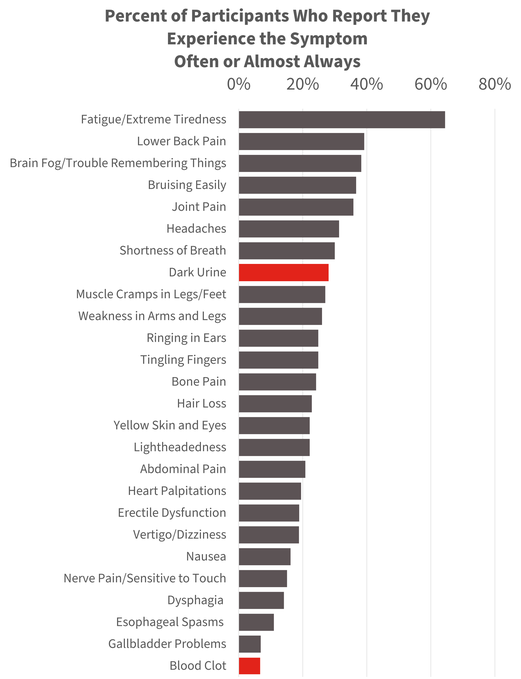
Analytic Methods

Includes data entered as part of an initial submission for each survey between April 2021 and April 2024

Quality of life is measured using PROMIS® tools which were scored and transformed to standardized t-scores.

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

RESULTS



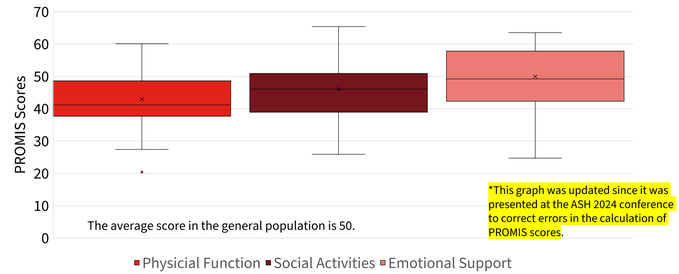
Bars in red are considered hallmark symptoms of PNH.

- **173** individuals with PNH consented to the registry and provided data during this time
- Study population reflects demographics typically seen in internet-based research
- Study participants report experiencing considerable symptom burden, especially fatigue, pain (lower back, head and joint), and brain fog
- Individuals with PNH are reporting impacts on their quality of life. Scores for participation in social activities and emotional support are comparable with the general population. For Anxiety and Pain Interferences, more than half of the study participants reported scores indicating some impairment.

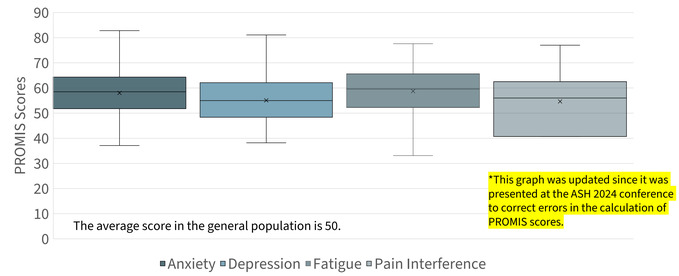
Study Participant Characteristics	Percentage
Demographic Characteristics	
Adult	99%
Female	65%
United States Resident	69%
Asian	12%
African American / Black	6%
Member of LGBTQ+ community	11%
No Post Secondary Education	16%
Unable to work	14%
Income below $25,000	12%
Diagnosis	
Average age of symptom onset	34
Average age at diagnosis	36
Co-morbid Conditions	
Aplastic Anemia	38%
Myelodysplastic Syndrome	9%
Health Care Utilization (6 months before first data entry)	
10+ PNH doctor visits	42%
PNH Hospitalization	26%
Treatments Received (ever at first data entry)	
Complement Factor Drugs*	68%
Eculizumab	45%
Ravulizumab	34%
Pegcetacoplan	9%
Blood Thinners	10%
Immunosuppressants	10%
Blood or Platelet Transfusion	10%

*Iptacopan and Danicopan were FDA-approved too recently to be captured in the registry during this initial 3-year period of data collection. Crovalimab was FDA-approved after this data collection period.

Distribution of PROMIS Measures Scores where **lower** scores indicate more impairment*



Distribution of PROMIS Measures Scores where **higher** scores indicate more impairment*



CONCLUSIONS

The Global PNH Patient Registry highlights the power and potential of patient-reported data to provide meaningful insights on the natural history of PNH in a clinically and demographically diverse population.

Plans include:

- ✓ Expanding recruitment and retention efforts
- ✓ Translating the registry into Spanish
- ✓ Examining the longitudinal data
- ✓ Developing processes to broaden the use of the data within the community

Considering the evolving landscape of anti-complement therapeutic options available for patients, we believe this registry will be invaluable for monitoring the experience of individuals living with PNH in an increasingly complex field.

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.