Characteristics of Patients Enrolled in a Pharmacist Managed Rare Disease Program: Highlighting National Diversity and Representation

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BACKGROUND

- Rare diseases affect millions of patients, but their low prevalence often leads to fragmented care, delayed diagnosis, and disparities in access to specialized therapies.[1]
- Specialty pharmacy programs for rare disease management offer patient-centered services such as tailored education and care coordination. These programs enhance outcomes and extend outreach to a diverse patient population nationwide.[2] By bridging the gap between patients and healthcare providers, they ensure timely access to essential treatments, and support research and development efforts which fosters innovation in rare disease therapies.

OBJECTIVE

- To characterize patients enrolled Pharmacist-Led Rare Disease Program (Ph-RDP), highlighting the diversity of the patients and the Program's national reach.

METHODS

- This study included patients from a large national specialty pharmacy with rare disease conditions as sleep disorder, sickle cell disease, x-linked hypophosphatemia, long chain fatty acid oxidation disorders (LCFAOD) with dispense for pitolisant, voxelotor, burosumab, l-glutamine and triheptanoin from CVS Pharmacy and enrolled in Ph-RDP between 3/15/2022 and 7/31/2024.
- Characteristics assessed included age, sex, rare disease type, geographic region, rural-urban residency, Social Vulnerability Index (SVI) quartile and drug treatment. Program engagement metrics included duration of enrollment and the number of completed assessments.
- Descriptive statistics included patient sociodemographic and clinical characteristics. Continuous variables were summarized with non-missing counts (n), mean, standard deviation (SD), median, interquartile ranges (IQR), minimum (min) and maximum (max). For categorical variables, counts and percentages were summarized.

RESULTS

- Among the 7,633 patients enrolled in the Ph-RDP, 55% were diagnosed with sleep disorders, primarily treated with pitolisant (**Figure 1**), while 34% had sickle cell disease, with voxelotor being the most common medication (**Figure 2**).

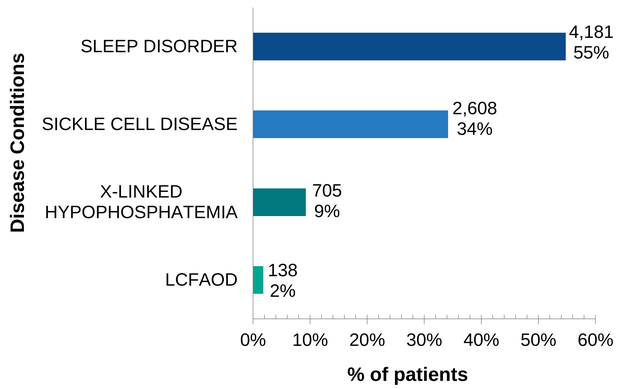

Figure 1. Proportions of patients by top rare disease conditions among Ph-RDP patients

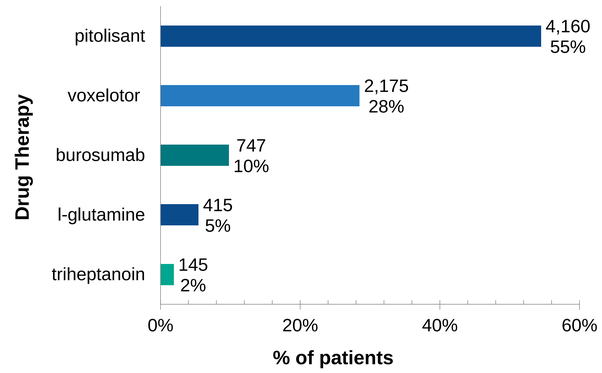

Figure 2. Proportions of patients by top drug therapy among Ph-RDP patients

- The program demonstrated high engagement, with 49% of patients (N=3,718) completing at least five assessments (**Figure 3**) and 48% (N=3,656) continuing in the program for over 200 days (**Figure 4**).

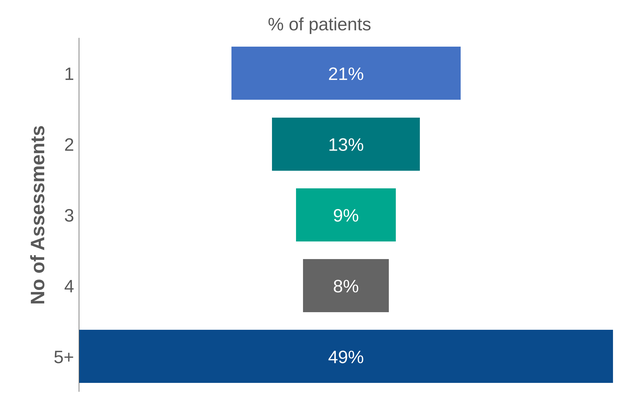

Figure 3. Proportions of patients by number of assessments

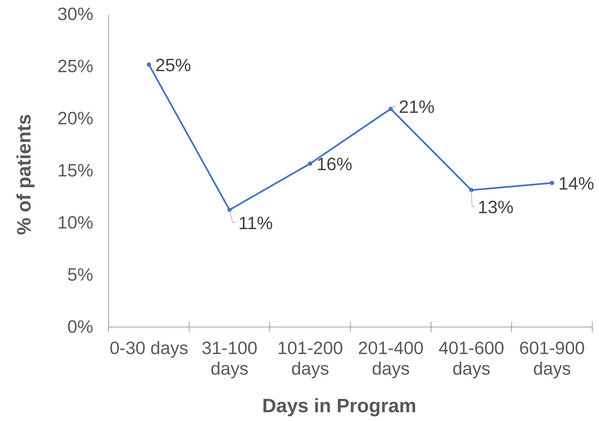

Figure 4. Proportions of patients by days in program

- The patient population was predominantly female (65%, n=4,934)) with mean age of 34 years (Figure 5 & 6), and a geographically diverse national representation: South (47%), Midwest (22%), Northeast (18%), and West (13%) (Figure 7). SVI quartile distribution showed 16% (n=1,203) of the patients in the most vulnerable quartile (Figure 8), average SVI was 0.48. Notably, 44% (n=3,358) of patients were in rural areas and 30% (n=2,253) in urban areas (**Figure 9**).

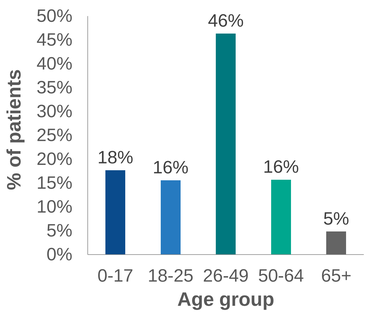

Figure 5. Proportions of patients by age group

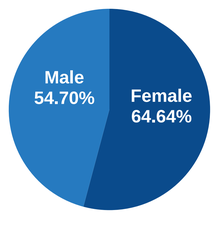

Figure 6. Proportions of patients by sex

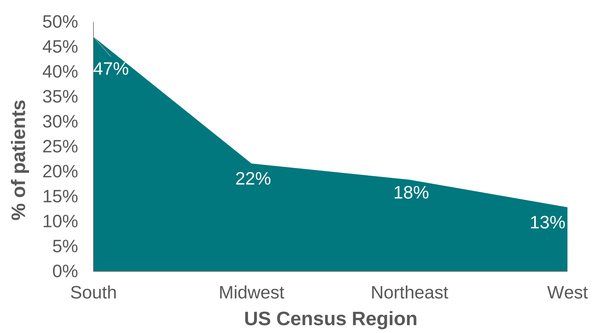

Figure 7. Proportions of patients by region

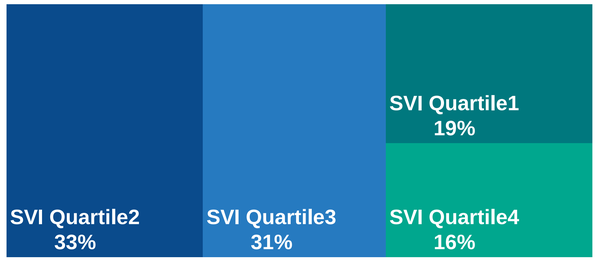

Figure 8. Proportions of patients by SVI Quartiles

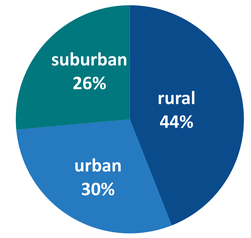

Figure 9. Proportions of patients by location

CONCLUSIONS

- The findings highlight the reach of a specialty pharmacy program, including substantial patient enrollment from rural, urban, and high vulnerability areas, serving as a proxy for providing accessible care to underserved populations.
- Patient participation and high engagement levels, evidenced by the number of assessments and duration of enrollment, emphasize the potential impact of the program to improve care and health outcomes.
- The large, national representative sample of patients with PAH offers opportunities for patient recruitment in clinical trials as well as in health economics and outcomes research.

REFERENCES

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