

# Pompe Community Members Face 2.5x Higher Risk of Mental Health Issues Compared to the General Population

Heather Shorten <sup>1</sup>, Nadia Bodkin <sup>2</sup>, JC Muyl <sup>3</sup>, and Heather Flaherty <sup>4</sup>

<sup>1</sup> Pompe Alliance, <sup>2</sup> Rare Advocacy Movement (RAM), <sup>3</sup> MyRareData, and <sup>4</sup> Level5 Insights

## Introduction

Mental health challenges are a pervasive yet often overlooked aspect of people living with rare diseases. For individuals with Pompe disease, a rare genetic disorder that affects muscle function and respiratory health, the burden extends beyond physical symptoms to significant emotional and psychological stress. The uncertainty of disease progression, the burden of managing complex symptoms, and the challenges in accessing specialized care contribute to elevated stress, anxiety, and depression among individuals living with Pompe disease and their caregivers. However, despite the clear need for mental health support, access to appropriate resources remains limited.

By investigating these challenges, this research seeks to underscore the urgent need for improved integration of mental health support within the Pompe disease community.

## Method

This study employed an online survey to evaluate the mental health experiences of individuals within the Pompe disease community. A total of 44 respondents participated in a questionnaire administered by the Pompe Alliance in collaboration with the Rare Advocacy Movement (RAM) and My RareData between Q3 2023 and Q3 2024.

The sample comprised 16 individuals diagnosed with Pompe disease and 28 caregivers of children or adults living with Pompe disease. The survey explored several aspects of mental health, including whether participants had received a mental health diagnosis, were seeing or had seen a therapist, whether a mental health professional was recommended at the time of diagnosis, satisfaction with mental health care, as well as information related to disease diagnosis and demographic details.

Additionally, qualitative data were collected through 18 one-hour interviews (n=9 patients, n=9 caregivers) conducted by Level 5 Insights, offering deeper insights into the emotional and psychological experiences of the Pompe community.

## Results and Discussion

### Prevalence of Mental Health Diagnoses:

- ▶ 57% of all respondents reported receiving a mental health diagnosis at some point, which is 2.5 times higher than the general population prevalence (22.8%, [source: NIH](#)).
- ▶ Common diagnoses among respondents included anxiety (40%), depression (27%), and PTSD (13%).

### Suicidal Ideation:

- ▶ 25% of all respondents have experienced suicidal thoughts, with the figure rising to 31% among patients. This is nearly double the prevalence of suicidal ideation in the general population (13.5%, [source: JAMA Psychiatry](#)).

### Access to Mental Health Services:

- ▶ Only 19% of patients who participated in this study were recommended to see a mental health professional at the time of diagnosis.
- ▶ 48% of all respondents have seen or are currently seeing a mental health professional. Reasons cited in the qualitative study for not seeking therapy include stigma around mental health, prioritizing physical health or other healthcare appointments and reliance on social support networks (e.g., family, friends, church).

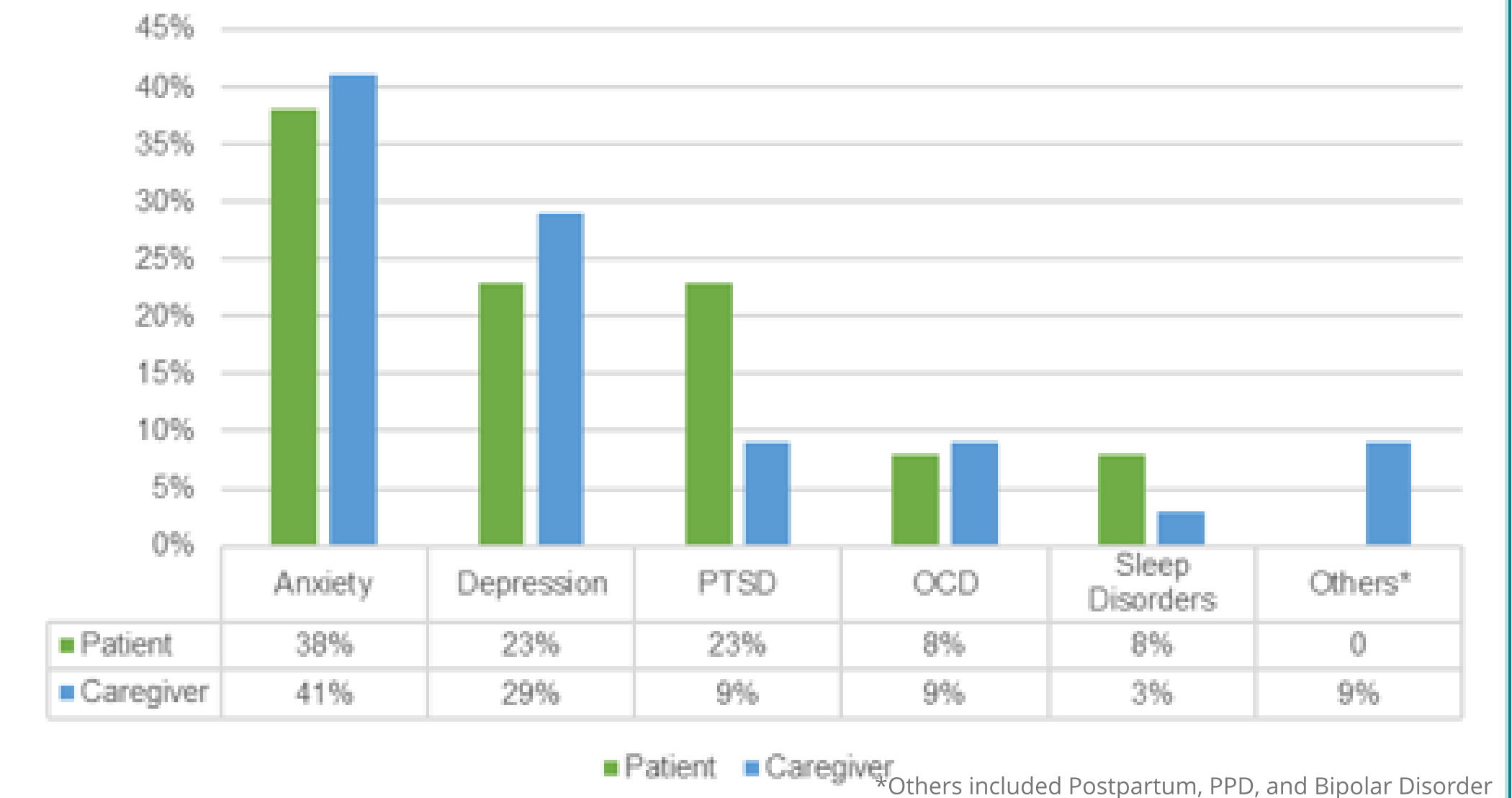
### Barriers to Seeking Therapy:

- ▶ Among the 52% who have never seen a mental health professional, 74% expressed a desire to do so. Barriers identified in the qualitative study include difficulty finding a local mental health professional for in-person therapy, lack of support from spouses or family members in pursuing therapy or difficulty finding a faith-based therapist or one who shares their values.

### Effectiveness of Mental Health Support:

- ▶ Among those diagnosed with Pompe disease who have accessed mental health support, 42% report that they found it helpful. This finding prompted a follow-up study to explore ways to enhance the effectiveness of mental health support for the Pompe disease community.

Mental Health Diagnoses Comparison: Patients vs. Caregivers



## Conclusion

This study reveals significant mental health challenges in the Pompe disease community, with a high prevalence of mental health diagnosis compared to the general population, low referral rates and barriers to seeking therapy. Less than half of those who received mental health support found it beneficial, highlighting the need to improve mental health support when available. These findings underscore the importance of developing strategies to improve both access to and also the quality of mental health care for individuals affected by Pompe disease.

The insights gained from this study lay the groundwork for further investigation and development of strategies aimed at improving mental health support for individuals affected by Pompe disease.

## Contact

**Website:** [www.rareadvocacymovement.com/mentalhealthforrare](http://www.rareadvocacymovement.com/mentalhealthforrare)

**Email:** [admin@rare360.life](mailto:admin@rare360.life)