

Mental Health Implications in Lysosomal Storage Diseases: Insights from the UK LSD Collaborative Community Survey

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INTRODUCTION

Lysosomal storage diseases (LSDs) are inherited metabolic disorders characterized by the accumulation of substrates within cells due to defective lysosomal functioning, leading to organ dysfunction and significant morbidity and mortality (Rajkumar, 2023). While the physical manifestations of LSDs are well-documented, recent insights highlight their profound impact on mental health. Pará et al. (2020) found that approximately two-thirds of LSD patients experience neurological issues, including psychiatric problems.

To delve deeper into this aspect, the UK LSD Collaborative, a coalition of seven UK-based patient advocacy groups, conducts the LSD Collaborative Community Survey biennially. This nationwide survey, open to individuals affected by LSDs, their families, and caregivers, explores various topics, including diagnosis, treatment access, and mental health.

METHODS

The survey was open to UK patients affected by all LSDs, plus their family members and carers. The survey was made available via the Survey Monkey platform and consisted of 75 questions.

It was distributed by The UK LSD Collaborative advocacy groups to their members and by LSD clinicians to their patients from 18 centres in the UK for both adult and pediatric patients. The survey took on average each respondent approximately 20 minutes to complete.

RESULTS

In total 325 people completed the survey all respondents were based in the UK. Key results from some questions are shown below.

Figure 1. Top 3 activities that have a positive impact on mental well-being

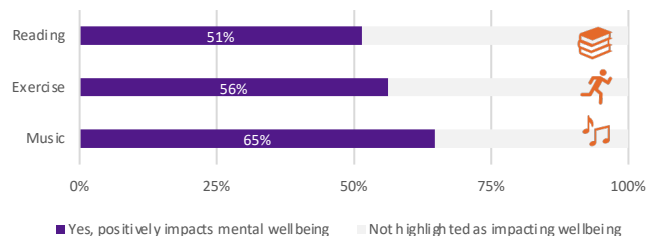


Figure 2. “Can you describe some of the key emotions you have experienced?” question WordCloud



CONCLUSION

Findings from the survey reveal that 30% of respondents report experiencing poor mental health, with only 28% receiving some form of counseling or mental health support, highlighting a crucial gap in care provision. Respondents cited anxiety, worry, anger, fear and loneliness as key drivers of their mental health challenges. Recognizing the potential for complementary approaches, the survey indicates that 65% of respondents find music beneficial for mental well-being, whether through playing or listening, while 56% endorse the positive effects of exercise. Integrating such non-pharmacological interventions alongside conventional treatments could significantly improve outcomes and enhance quality of life for LSD patients and caregivers.

These outcomes underscore the importance of a holistic approach to mental health care in LSDs, emphasizing collaboration among clinicians to integrate diverse therapeutic modalities into patient management, while empowering healthcare professionals who treat rare disease patients and integrating mental health support into rare disease services (Spencer-Tansley et al., 2022).

REFERENCES

- Pará, C., Bose, P., & Pshezhetsky, A. V. (2020). Neuropathophysiology of Lysosomal Storage Diseases: Synaptic Dysfunction as a Starting Point for Disease Progression. *Journal of Clinical Medicine*, 9(3), 616. doi: 10.3390/jcm9030616. PMID: 32106459; PMCID: PMC7141115.
- Rajkumar, V., & Dumpa, V. (2024, July 24). Lysosomal Storage Disease. In *StatPearls*. Treasure Island (FL): StatPearls Publishing. Retrieved from <https://www.ncbi.nlm.nih.gov/books/NBK563270/>.
- Spencer-Tansley, R., Meade, N., Ali, F., et al. (2022). Mental health care for rare disease in the UK – recommendations from a quantitative survey and multi-stakeholder workshop. *BMC Health Services Research*, 22, 648. doi: 10.1186/s12913-022-08060-9.