

CLUSTER ANALYSIS OF PATIENT-REPORTED OUTCOME MEASURES OF ADULTS WITH X-LINKED HYPOPHOSPHATAEMIA

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Objective

We set out to identify distinct subgroups of adults with X-linked hypophosphataemia (XLH) based on their patient-reported outcomes.

Material and Methods

We used data from adults with XLH registered in RUDY, a UK rare diseases cohort study. We applied cluster analysis using the K-means method to seven patient-reported outcome measures (PROMs): EQ5D-5L and SF-36 physical and mental component scores (PCS and MCS) as measures of quality of life (QoL), FACIT-F of fatigue, PainDETECT and SF-MPQ-2 of pain, PSQI of sleep quality, and HADS of anxiety and depression. Instruments' scores were standardised so that all variables would be comparable. The average silhouette and elbow method were used to determine the optimal number of clusters.

Results

17 participants (76.5% females, median age 41 years) completed all seven PROMs at their first submission. Optimal number of clusters was two.

The "higher-score" cluster included 11 participants reporting higher (better) mean QoL scores compared to the "lower-score" group (EQ5D-5L=0.817, PCS=45.40 and MCS=51.31, compared to 0.288, 19.54, and 43.44, respectively). They also reported better sleep quality (PSQI=7.64 compared to 12.67), less fatigue (FACIT-F=125.32 vs 78.00), less pain (SF-MPQ-2=1.03 compared to 3.58), and lower likelihood of being categorised with neuropathic pain (PainDETECT=5.64 vs 14.17) and depression (HADS=3.18 vs 8.17). Anxiety was found to be similar between clusters (HADS=6.73 vs 6.50).

Conclusions

This analysis shows that adults diagnosed with XLH report broadly heterogeneous PROMs which, even with a limited sample size, allow them to be split into two very distinct subgroups, one with consistently better pain, sleep, fatigue, and depression than the other. Comparisons with other rare disease groups, other musculoskeletal conditions, and the general population would provide an important benchmark.

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Disclosures

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