A cross-sectional study of patient-reported outcomes of individuals with X-linked hypophosphataemia within the UK

Sophie Cole, Maria T. Sanchez-Santos, Joe Barrett, Nathanael Gray, Alison Turner, Spyros Kolovos, M. Kassim Javaid, Rafael Pinedo-Villanueva
University of Oxford, Oxford, United Kingdom

Abstract

Objective

We described the patient-reported outcomes (PROs) of adults with X-linked hypophosphataemia (XLH) and examined if they are correlated.

Methods

Data were obtained from the RUDY study, which collects information from participants with rare diseases in the UK. Participant responses to nine instruments were extracted: EQ5D-5L and SF36 physical (PCS) and mental component score (MCS) as measures of quality of life, ESS and PSQI of sleep quality, PainDETECT and SF-MPQ-2 of pain, FACIT-F and FSS of fatigue, and HADS of depression and anxiety. Differences in mean scores between age groups and gender for first submitted instruments were tested using ANOVA and independent samples t-tests, respectively, and correlation between instruments examined via repeated measures correlation.

Results

The sample was comprised of 48 participants with XLH (77% females, median age 46 years). Mean scores were: EQ5D-5L=0.65, SF36-PCS=32.7, SF36-MCS=48.4, ESS=5.9 and PSQI=8.9, FSS=32.8 and FACIT-F=104.4, HADS-depression=4.7 and HADS-anxiety =6.2, SF-MPQ-2=1.9 and PainDETECT=9.3. Severe or extreme problems were reported in all instruments, with mobility (23%) and pain (23%) from EQ5D, and functional wellbeing (mean=18.2) and fatigue (mean=31.3) in FACIT-F being the most frequent.

There was no statistically significant difference in mean scores by age group, or by gender except for PSQI, indicating that females (mean=9.6) experience a slightly poorer quality of sleep than males (mean=6.4).

We found low to moderate correlation between instruments, few being statistically significant. The lowest correlation coefficient was between SF36-MCS and PainDETECT (r=-0.019) and highest between PSQI and FSS (r=0.579). The highest statistically significant correlations ($p<0.05$) were between SF36-MCS and FACIT-F (r=0.513).

Conclusions

People with XLH reported most severe problems in mobility, pain, functional well-being and fatigue. Mental health and fatigue reported the highest correlation. More research is needed to identify the PROMs that most consistently capture the main health problems and changes experienced by people with XLH.

Acknowledgement

We thank RUDY participants.