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PHYSICAL FUNCTION AND MOBILITY IN ADULTS WITH X-LINKED HYPOPHOSPHATEMIA

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Objective: X-linked hypophosphatemia (XLH) is a rare genetic disorder affecting phosphate metabolism. Whilst muscle weakness has been reported in adults with XLH, there is little data describing detailed physical function. We therefore examined upper and lower limb function and aerobic fitness in UK adults with XLH and assessed the relationships between physical function and mobility.

Methods: Adults with XLH were recruited as part of an ongoing UK-based prospective cohort study, the Rare and Undiagnosed Diseases Study (RUDY). Participants underwent a clinical visit and physical examination. This included grip strength and jump power assessed by mechanography, 6-min walk test (6MWT) and short physical performance battery (SPPB). Scores were compared with existing age and sex-specific normative data using t-test, whereas correlations among outcomes were processed using Pearson's correlation coefficient.

Results: Fifteen adults with XLH (9 males and 7 females), with a mean age of 47.3±16.7 y were enrolled to the study. Grip strength was 26% lower and jump power 57% lower in individuals with XLH than normative values, with greater deficits evident in the lower than upper body (all $p<0.01$). Aerobic fitness was 40% lower in XLH individuals when compared to reference values ($p<0.001$). Mean SPPB score was 9.0±3.2, with 5/15 individuals having a score of <10 indicating impaired mobility. Univariate correlations revealed that age ($r=-0.635$, $p=0.011$) handgrip strength ($r=0.672$, $p=0.006$), jump power ($r=0.651$, $p=0.016$) and aerobic fitness ($r=0.874$, $p=0.001$) were all highly correlated to mobility as measured by SPPB.

Conclusion: Adults with XLH had weaker lower body power appears than other components of physical function. Upper and lower limb muscle function and aerobic fitness were all strongly associated with impaired mobility in this population, which suggests that the origin of mobility deficits may be multifactorial. Further studies are required to understand underlying mechanisms, and to develop novel treatment approaches to improve physical function and mobility.