Physical function and mobility in adults with X-linked hypophosphatemia

Giorgio Orlando[1], Shane Clark[2], Kassim M. Javaid[2], Alex Ireland[1]

[1]Research Centre for Musculoskeletal Science & Sports Medicine, Manchester Metropolitan University, UK
[2]Department of Rheumatology, University Hospitals Bristol, Bristol, UK
[3]Nuffield Department of Orthopaedics, Rheumatology and Musculoskeletal Sciences (NDORMS), University of Oxford, UK

Manchester Metropolitan University

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Introduction

X-linked hypophosphatemia (XLH) is a rare genetic skeletal disorder affecting phosphate metabolism. Whilst muscle weakness has been reported in adults with XLH, there is little data describing detailed physical function.

Aims:

I. to examine upper and lower limb function and aerobic fitness in UK adults with XLH;

II. to assess the relationships between the different components of 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 study).

Participants underwent a physical examination to assess:

I. handgrip strength (isometric dynamometer);

II. jump power (jumping mechanography);

III. aerobic fitness (six-minute walk test, 6MWT);

IV. mobility (short physical performance battery, SPPB).

Scores were compared with normative values using t-test, whereas univariate correlations were processed using Pearson's correlation coefficient.

Results

The cohort included 21 adults with XLH —10 males and 11 females— with a mean age of 44.2±16.9 years and a BMI of 27.6±4.4.

Grip strength (Fig. 1) was 22% lower (p=0.016) and jump power 59% lower in individuals with XLH than normative values (p<0.0001), with greater deficits evident in the lower than upper body (p=0.01).

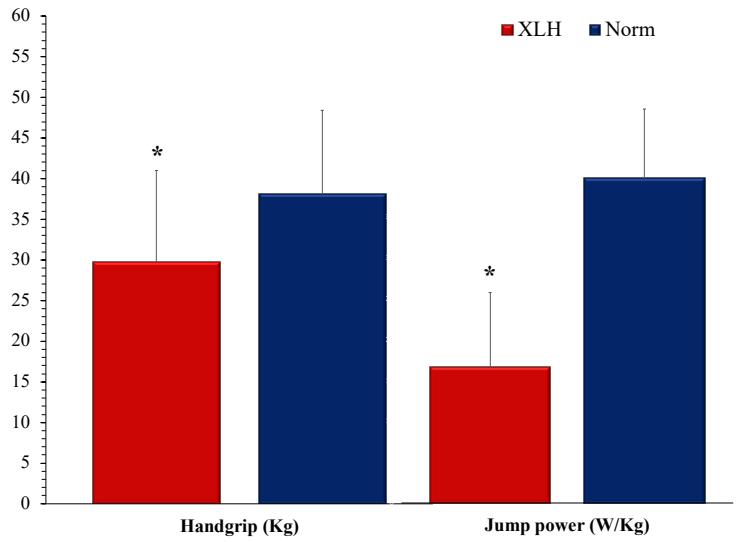

Figure 1. Upper and lower body function in people with XLH compared with normative values.

Aerobic fitness (6MWT, Fig. 2) was 40% lower in XLH individuals when compared to reference values (p<0.001).

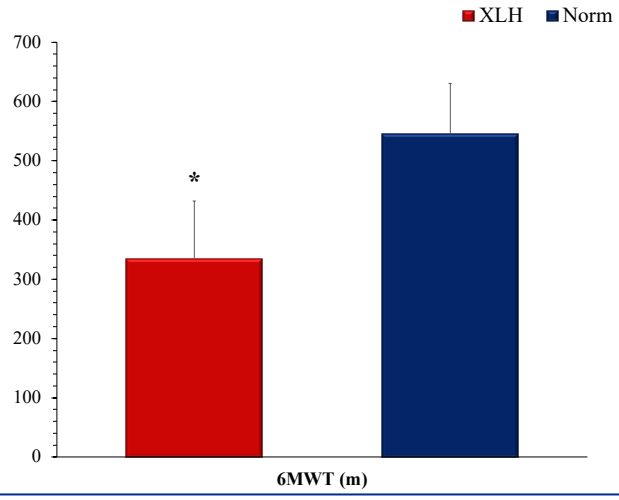

Figure 2. Aerobic fitness in adults with XLH and the general population.

Mean SPPB score was 9.0±3.2, with 5 individuals having a score of <10 indicating impaired mobility.

Univariate correlations (Table 1) revealed that age, handgrip strength, jump power and aerobic fitness were all highly correlated to mobility (SPPB).

	Univariate Analysis	
Variables	Rho	*p values*
Age	-0.666	0.001
Handgrip strength	0.661	0.001
Jump power	0.645	0.005
6MWT	0.845	<0.001

Table 1. Pearson correlation between SPPB and upper and lower limb function and aerobic fitness.

Conclusions

- Muscle strength, power and aerobic fitness are impaired in adults with XLH.
- Lower body function appears more affected than other components of physical function.
- Muscle function and especially aerobic fitness are strongly associated with impaired mobility.

Specific countermeasures should be included in the standard care of adults with XLH to prevent and counteract the loss of mobility.

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Email: G.Orlando@mmu.ac.uk Twitter: @GioOrl1986