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Characterising spillover effects and carer burden in X-linked hypophosphataemia

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KEY FINDINGS

Qualitative findings indicate that carers and family members provide considerable support for adults with XLH resulting in a broad range of impacts to carers and family members' lives and highlight areas of carer-burden not well captured by the EQ-5D.

INTRODUCTION AND OBJECTIVE

- Health-related quality of life (HRQoL) impacts of disease extend beyond the patient.
- 'Spillover effects' are the HRQoL impacts experienced by any individual in contact with a patient - carers, family members, partners and friends.
- X-linked hypophosphataemia (XLH) is a chronic rare metabolic bone disease, with a high disease burden (1,2).
- XLH is genetic and in many cases hereditary, with carers of XLH patients often being patients themselves.
- To date, no research has explored the possible spillover effects of XLH.

OBJECTIVE: To describe the spillover effects of XLH and measure their impacts on carers and family members of adults with XLH.

METHODS

- Informal carers and family members of adults with XLH completed the study and were recruited through XLH UK, the UK patient organisation supporting people with XLH.
- All participants provided informed consent. The study protocol was reviewed by an Independent Review Board.
- Quantitative data collected included: (1) participants' and care recipients' demographic and clinical information; (2) the EQ-5D-5L; (3) the Work Productivity and Activity Impairment (WPAI) questionnaire (3).
- Qualitative data were collected via one-to-one semi-structured interviews conducted virtually (Via Zoom or telephone).

QUANTITATIVE FINDINGS

- Twenty informal carers or family members of adults with XLH took part in the study. Four participants had XLH themselves.
- The mean age of participants was 48.5 years (range 26-83) and the majority were male (N=13/20; 65%).
- The WPAI revealed an overall work impairment of 28% and activity impairment of 42%.
- The EQ-5D-5L demonstrated lower HRQoL for informal carers and family members of adults with XLH compared to the UK general population data.
- Informal carers with XLH (n=4) had much lower HRQoL compared to both informal carers without XLH (n=16) and the UK general population.
- Informal carers without XLH had HRQoL only slightly less than that of the UK general population. **(Figure 1)**

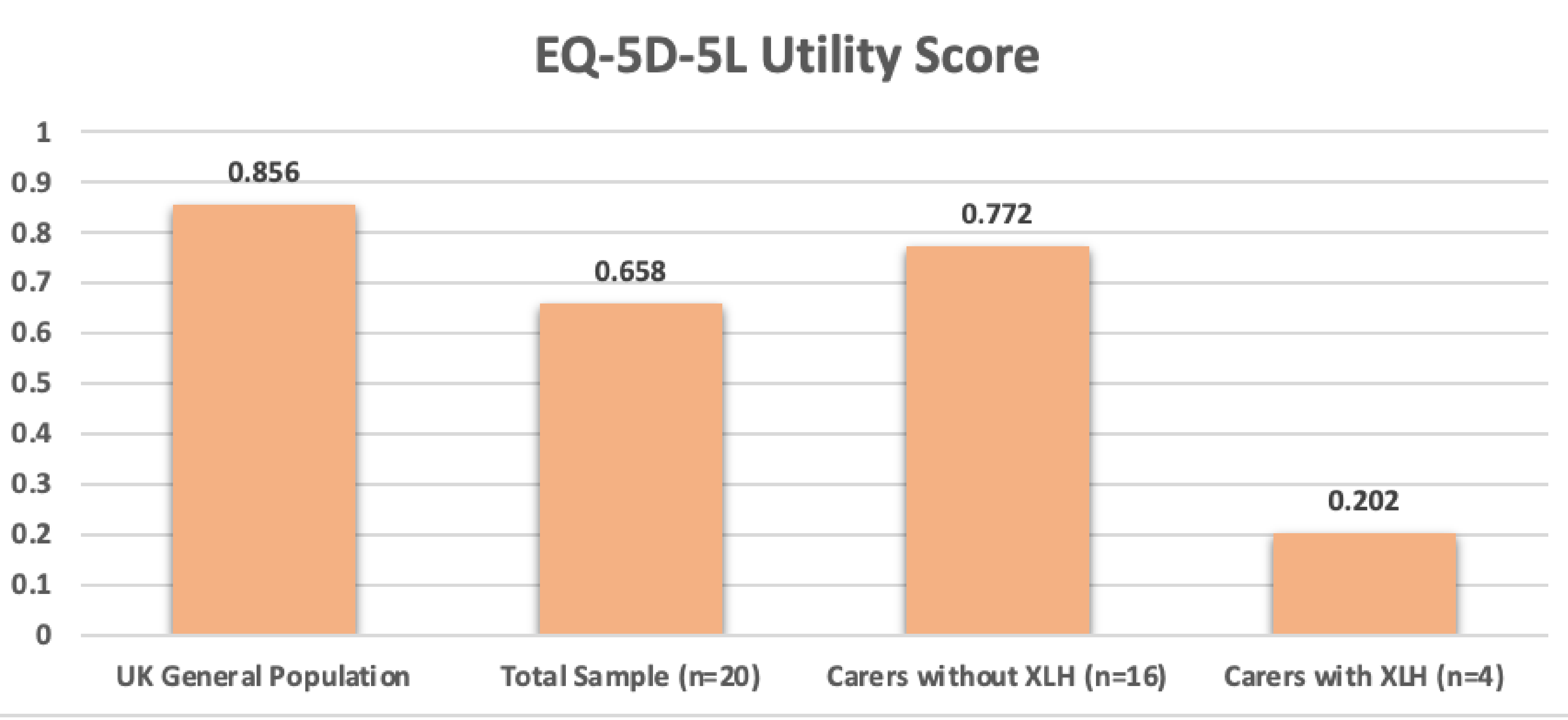

Figure 1. EQ-5D-5L Utility scores of participants versus UK General Population

QUALITATIVE FINDINGS

- Informal carers and family members provide adults with XLH support in several areas.
- Providing support for adults with XLH resulted in spillover effects on participants' HRQoL, with impacts greatest among informal carers with XLH **(Figure 2).**
- Although on the EQ-5D-5L most carers without XLH reported 'no problems' on the usual activities, and, 'no problems' or 'slight problems' on the anxiety and depression domains, almost all participants qualitatively described substantial impacts to their daily activities and their emotional wellbeing.

Figure 2. Health-related quality of life impacts experienced by carers and family members of XLH patients

Impacts on family members and carers		
Emotional wellbeing	Participants reported experiencing worry, stress and guilt. Carers with XLH expressed feeling guilty 'passing on' XLH to their adult child and noted the added burden of worrying about their own health.	***"It's not nice, watching your partner slowly deteriorate… It does have an impact on you, It's hard to put it into words" (PPT15)***
Day-to-day activities	Participants described having no time or freedom left to engage in activities they would have otherwise liked to participate in.	***"Sometimes I don't know who I am anymore … the little things that I used to do like listen to music and things like that … I don't really get a chance to do anymore" (PPT09)***
Work	Participants reported having increased or decreased working hours, or having to work in unpreferred ways. Some reported having to stop work altogether to provide support.	***"I was working nights, which made our life easier because it meant I was home during the day and then come [adult with XLH's] bedtime I was at work … It takes quite a big toll on your lifestyle" (PP02)***
Financial impacts	Participants reported impacts of a reduced income and the financial burden of medical and transport costs. For some, this meant financial instability.	***"I get carer's allowance. I kind of gave up all financial stability to look after [them]" (PPT06)***
Social and relationship impacts	The ability to socialise with friends and other family members was limited for some. Positive and negative relationship impacts between the participant and the adult with XLH were reported.	***"I just feel guilty being able to do something that I know [they] might enjoy … so therefore I decline it. I don't like going to things that my partner can't go and enjoy or doing something without [them]" (PPT15)***
Physical impacts	Participants described experiencing body aches, sleeplessness, and tiredness. For carers with XLH, physical impacts were amplified due to their own XLH diagnosis.	***"Physically it makes it more difficult because I'm limited on what I can do quite a lot because of the pain, or if I'm on days where I haven't got a lot of energy it can make it quite difficult" (PPT20)***

CONCLUSIONS

- This study highlights the substantial spillover effects of XLH.
- The disparity between the EQ-5D utility data and the substantial burden reported in the qualitative data for carers without XLH in the domains of usual activities, and, anxiety and depression, raises important questions about the appropriateness of the EQ-5D to measure spillover effects in XLH and other disease areas.

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