Development of a framework for a networked service model for the care of adults with rare bone conditions: healthcare professional survey for X-linked hypophosphataemia (XLH)

HSD9

GPR Clunie,[1] MK Javaid,[2] O Gardiner,[3] V Hayes,[4] L Mathieson,[4] R Keen[5]

[1]Cambridge University Hospital Trust, Addenbrookes, UK; [2]Nuffield Department of Orthopaedics, Rheumatology and Musculoskeletal Sciences, University of Oxford, UK; [3]XLH UK, Kington, UK; [4]Kyowa Kirin International, Marlow, UK; [5]Royal National Orthopaedic Hospital, Stanmore, UK

Introduction

- X-linked hypophosphataemia (XLH) is a rare, genetic, progressive, phosphate-wasting bone disorder that starts in early childhood and causes skeletal morbidities, stiffness, pain and impaired physical function, which continue through the patient's life.
- Children with rare bone diseases in England benefit from multidisciplinary management whereas this is not currently the case for adults.
- A framework for a networked service model has been proposed for the care of adults in England with rare bone conditions, developed in collaboration with a steering committee of clinicians and patient group representatives and sponsored by Kyowa Kirin Ltd.
- The framework will be piloted at Addenbrookes Hospital in Cambridge then, if feasible and effective, rolled out across the Rare Disease Collaboration Network (RDCN) centres.
- Here, we report a survey of healthcare professionals to understand current access to, and quality of care, for adults with XLH, providing a baseline to assess the impact of the proposed networked service model.

Objective

To assess the existing provision of care for adults with rare bone diseases in England, prior to the roll out of the pilot programme across RDCN centres

Methods

- A steering committee comprising experts in rare metabolic bone diseases and representatives of adults living with XLH developed a survey to understand access to, and quality of, care for adults with XLH.
- Survey questions were linked to proposed key performance indicators in workforce and staffing capacity, service availability, care coordination and patient-centric care.
- The survey was sent by email to one clinician (rheumatology, endocrinology or metabolic disorders) at each of 22 RCDN centres.
- The survey was designed on and disseminated via Survey Monkey and was conducted between October 2023 and January 2024.

Results

- Responses were received from 11 centres in England and Scotland (response rate 50%).

Workforce and staffing

- Eight of the completed surveys (73%) confirmed that a named clinical lead for the service was highlighted on patient correspondence (**Figure 1**). Patients could contact the clinical lead by email or phone in all cases.
- Nine centres did not have a formal care coordinator to coordinate care across specialties (**Figure 2**).
- At the two centres that had a formal care coordinator, respondents gave scores of 4 and 5 to indicate whether the proportion of time dedicated to rare bone diseases by this coordinator was sufficient (1 not all sufficient; 10 very sufficient). Thus, capacity was a limiting factor.

Figure 1. Clinical lead named on patient correspondence (response rate 100%)

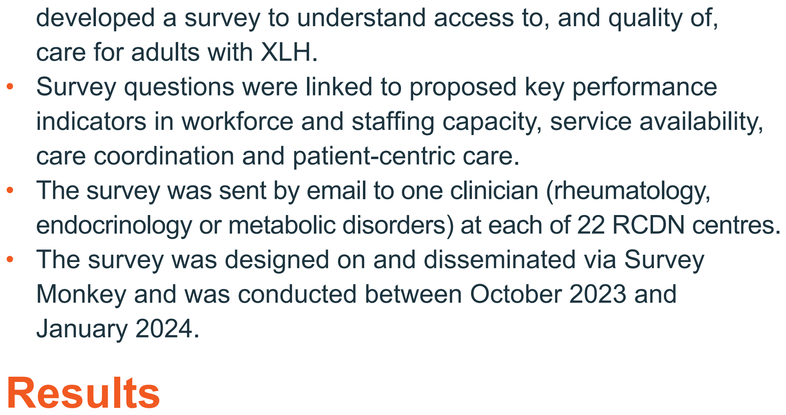
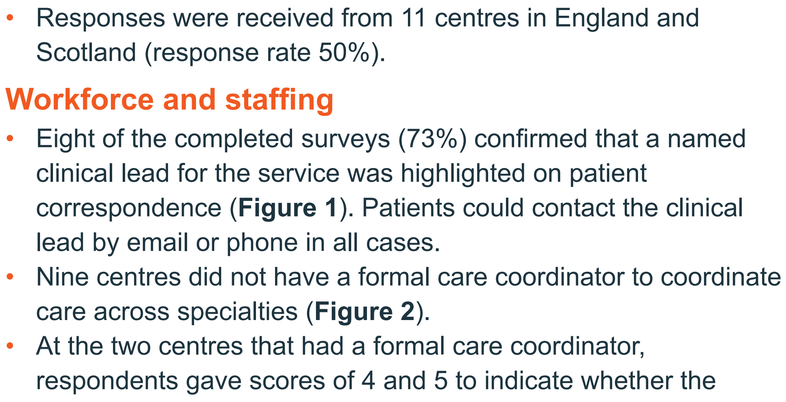


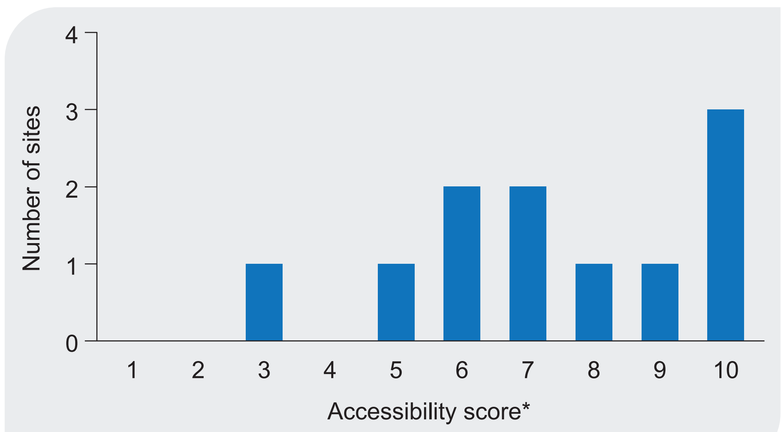
Figure 2. Formal care coordinator (response rate 100%)



Service availability

- All respondents confirmed that a genetic counselling and diagnostics service with a clinical genetics specialist was available within the hospital trust or regional network.
- Seven respondents (63%) rated the accessibility of the genetic counselling and diagnostics service as 7 or higher using a scale of 1–10 (not at all accessible to very accessible). Three of these gave a score of 10 (the most frequent response) (**Figure 3**).

Figure 3. Accessibility of genetic counselling and diagnostics service (response rate 100%)



*Scored from 1 (not at all) to 10 (very accessible)

Care coordination

- Eight respondents (73%) confirmed that a transitional care pathway with a named clinical lead was available (**Figure 4**).
- Nine respondents (82%) participated in multidisciplinary team (MDT) meetings (**Figure 5**).
  - Six centre leads had participated in at least two-thirds of MDT meetings in the last year but two had participated in only 10%.
  - The specialties involved in the MDT varied across centres (**Table 1**). Most (6/9) MDTs included genetics, radiology and endocrinology; five included a rheumatologist.
  - The number of specialties involved in MDTs ranges from two (3 centres) to seven (1 centre).
  - Respondents commented on the lack of resources available for MDT support (e.g. coordinators, administrators) and the difficulty of planning sufficient time for consultants across specialties to attend MDT meetings.

Figure 4. Transitional pathway with named clinical lead available (response rate 100%)

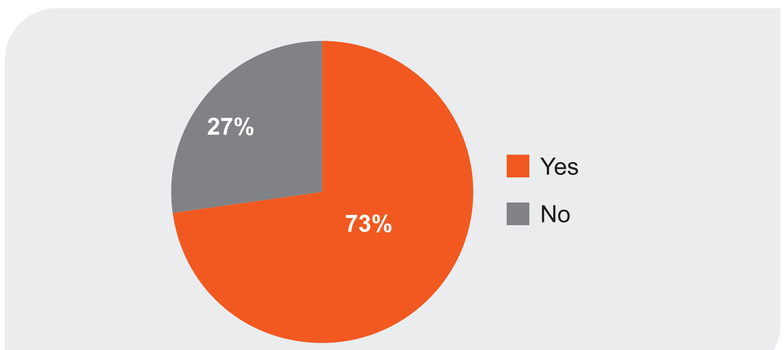
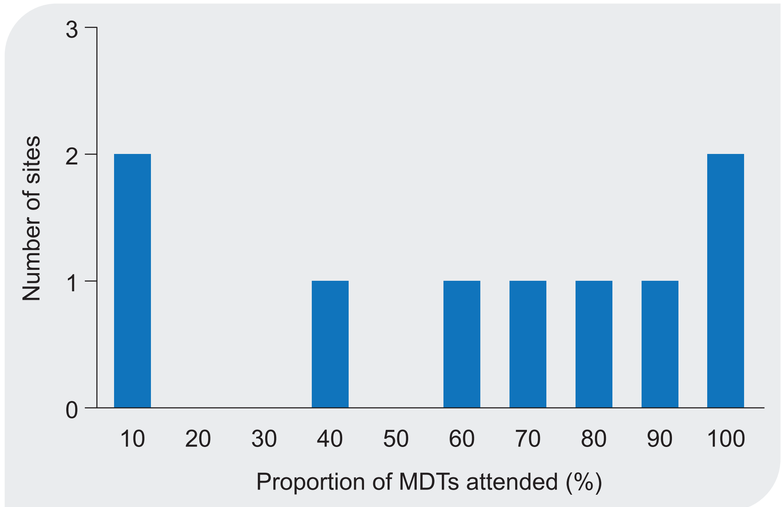


Figure 5. Participation in regional and/or national rare bone disease related multidisciplinary team (MDT) meetings in previous year (response rate 82%; 9/13)



Patient-centric care

- Ten respondents (91%) reported that a formal management template was not followed for all patients (**Figure 6**).
  - The management plan followed by one respondent included mental health and psychological support and regular follow-up but did not include physiotherapy, bone chemistry measurements, renal ultrasound or dental check-ups.
  - One commented that a clinical pathway would improve patient care.
  - One pointed out that it was not possible to refer patients directly to local dental or psychology services.

Table 1. Structure of MDTs: each column indicates the members of the MDT listed by respondents who participated in MDTs (n=9)

Rheumatology	✔	✔	✔	✔				✔	
Endocrinology		✔		✔	✔	✔		✔	✔
Metabolic medicine			✔						
Clinical biochemistry					✔				
Osteoporosis	✔								
Radiology	✔	✔		✔	✔	✔	✔		
Orthopaedics	✔	✔	✔						
Genetics	✔	✔	✔	✔		✔	✔		
Chemical pathology	✔								
Central nervous system	✔								
Paediatrics		✔	✔						
Geriatrics					✔				
Specialist nurses									✔
Number of specialties in MDT	7	6	5	4	4	3	2	2	2

- Two respondents reported that feedback surveys had been sent to patients living with rare bone diseases in the last year (one using the National Health Service Friends and Family Test).
  - Of the nine that did not send out surveys, two relied on clinical consultations and four reported lack of resources.
- Seven centres (64%) had rare bone condition service registries with basic demographic and diagnostic data (**Figure 7**).
- Nine centres (82%) directed patients to advocacy organizations providing support for patients with XLH.

Figure 6. Is formal management plan followed for all patients? (response rate 100%)

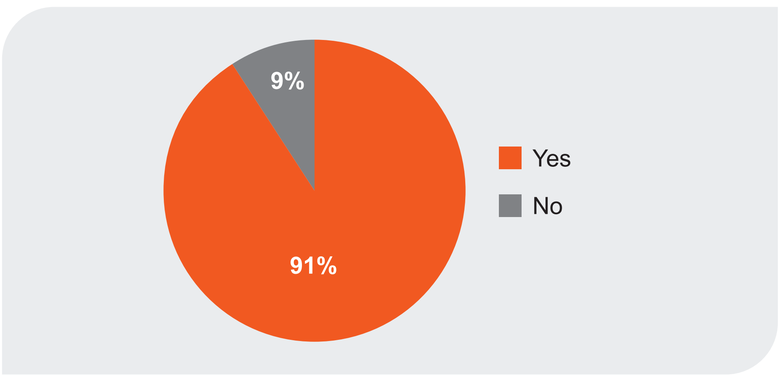
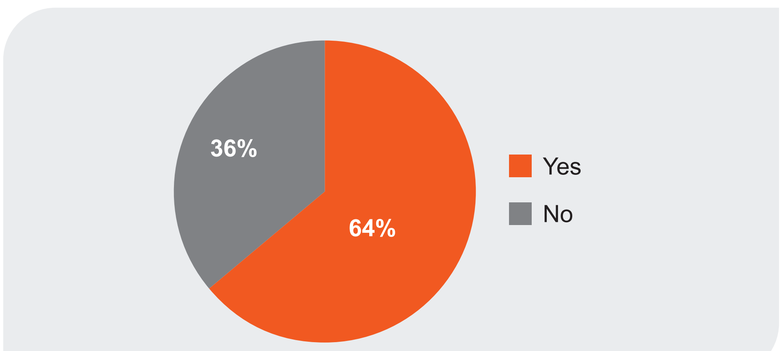


Figure 7. Rare bone condition service registry/database available (response rate 100%)



CONCLUSIONS

- **This survey highlights variability in healthcare resource and service infrastructure, a risk factor for inequality of healthcare provision.**
- **A national strategy to address this variation in service provision should be developed.**
- **The survey could be repeated annually to assess improvements and therefore the effectiveness of the pilot in improving patient care.**

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