Understanding XLH's Impact: Incorporating patient perspectives to develop a comprehensive conceptual disease model (CDM) to drive patient engagement and the generation of meaningful patient experience data

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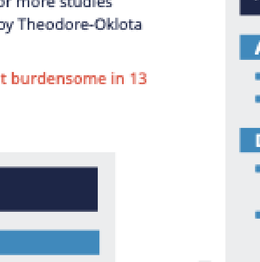
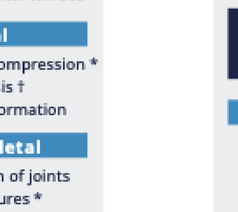
Patient-reported experiences of XLH can guide the selection of meaningful trial endpoints and ensure a patient-centric drug development programme

Living with XLH affects nearly every aspect of life, for both patients and their families.



Adults with XLH report significant impairments on quality of life.



Pain is reported as the most burdensome symptom for adults with XLH.

BACKGROUND & AIMS

- X-linked hypophosphatemia (XLH) is a rare genetic disorder with a prevalence of 3.9-5 in 100,000 live births.[1]
- A mutation in the phosphate-regulating gene causes elevated serum levels of fibroblast growth factor 23 (FGF23), limiting renal phosphate re-absorption resulting in hypophosphatemia.[1]
- XLH can cause musculoskeletal, neurological, auditory, and dental complications, and can have a significant impact on quality of life.[1]
- A 2018 study published a conceptual model summarizing concepts reported in interviews with patients with XLH in the United States.[2]
- The study presented here aimed to develop an updated CDM to synthesize important concepts from the literature more broadly.

CONCLUSIONS

- XLH affects nearly all body systems and has a significant impact on patients' quality of life.
- The draft CDM provides the foundation for a comprehensive clinical outcome assessment strategy to drive further patient-centred research.
- Subsequent qualitative research, such as concept elicitation interviews, will be conducted in the next phase of research to gain further insight into the experiences of people with XLH and refine the CDM.

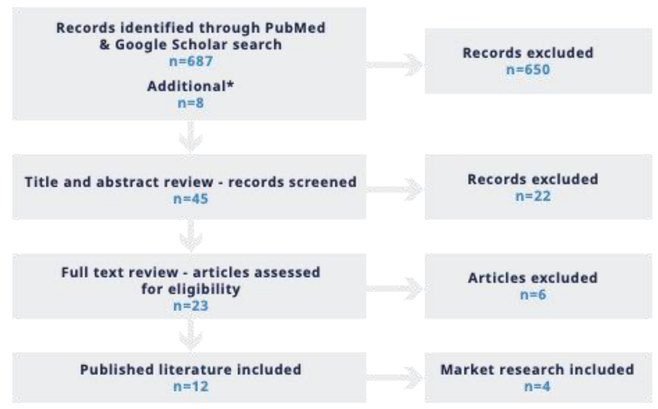
METHODS

- A targeted literature review (TLR) was conducted, prioritizing qualitative, patient experience data published since 2018.
- Due to the scarcity of recent qualitative studies, findings from gray literature, including market research insights, supplemented scientific data.
- Patient-reported signs, symptoms and impacts of XLH were extracted from the literature and synthesised in the revised CDM.

RESULTS

- 16 articles meeting eligibility criteria were included in the data extraction (Figure 1) and informed the development of the draft XLH CDM (Figure 2).
- The model summarizes published and unpublished literature, building on findings from the 2018 CDM[2].
- 29 signs and 34 symptoms were reported
  - Symptoms were often reported as progressive.
  - Chronic pain was reported as the most burdensome symptom and occurs in several areas of the body.
- XLH has widespread impacts on daily life, affecting emotional well-being, work, finances, physical functioning, relationships and sleep. The impacts of XLH also extend to the family.

Figure 1 PRISMA flow diagram for TLR



*Grey literature and market research

Figure 2 Draft conceptual disease model of XLH signs, symptoms and impacts in adults

ACKNOWLEDGEMENTS AND DISCLOSURES

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