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The burden of X-linked hypophosphatemia on patients and caregivers in South Korea: a narrative interview approach

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Background

- X-linked hypophosphatemia (XLH) is a rare genetic disease caused by mutations in the phosphate-regulating gene on the X chromosome and deteriorate the quality of life not only in patients but also in their caregivers.[1)]
- This study aims to investigate the poorly understood burden of the disease on patients with XLH and their caregivers in Korea.

Methods

Study design

- Face-to-face narrative interviews performed from November 2023 to February 2024.
- Participants were recruited via an open recruitment announcement through the Korean Organization for Rare Diseases (KORD) or snowball sampling.

Study population

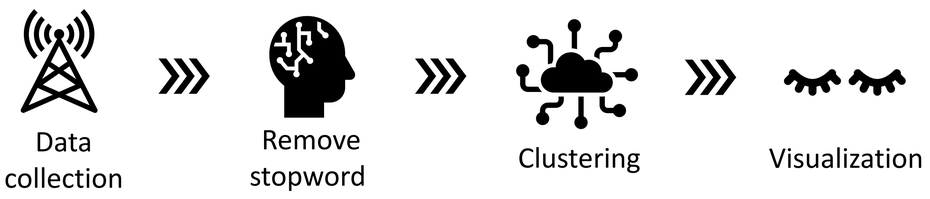
- Adults aged ≥19 years with a clinical diagnosis of XLH.
- Caregivers of patients <12 years with with a clinical diagnosis of XLH.

Outcome measurements

- Narrative disease burden explored with open-ended questions.
- Domains of burden of diseases in physical/psychological impacts, healthcare utilization, daily life discomfort, and life satisfaction.

Statistical analysis

- All the study variables were descriptively analyzed.
- Concept en was applied to obtain comprehensive insights into patient experlicitatioiences for qualitative analysis.



- Topic analysis was conducted using Microsoft Excel.
- Word cloud clustering was performed with Voyant Tools.

Discussions

- Patients with XLH and their caregivers experienced significant symptoms, financial burdens related to treatment, social impacts, and emotional challenges.[2, 3)]
- Most patients with XLH experienced pain. Treatment burdens were not only financial but also included the inconvenience of frequent drug administration and hospital visits.
- The psychological and emotional burdens of the disease were significant, with patients and caregivers reporting feelings of depression, frustration, worries about the future, and guilt.

Conclusions

- This study shed the light on the multifaceted disease burden experienced by XLH patients, revealing differences between adult patients and caregivers.
- It emphasizes the necessity for tailored management strategies to alleviate the diverse impacts of XLH on both patients and caregivers.

Acknowledgment

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References

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Results

Table 1. Demographics and clinical characteristics of XLH patients

Characteristics	Total (n=12)	Adult (n=4)	Pediatric (n=8)
Age (year), mean (SD)	16.25 (14.54)	34.75 (8.66)	7 (2.56)
Female, N (%)	9 (75.00)	4 (100.00)	5 (62.50)
Height (cm), mean (SD)	123.33 (20.73)	143.75 (11.95)	113.11 (16.01)
Weight (kg), mean (SD)	26.34 (14.20)	43 (6.63)	18.01 (7.74)
Age at symptom onset (year), mean (SD)	2.08 (1.51)	2.75 (1.71)	1.75 (1.39)
Age at XLH diagnosis (year), mean (SD)	3.17 (3.49)	5.75 (5.12)	1.88 (1.46)
History of XLH treatment *, N (%)			
Phosphate supplements	11 (91.67)	3 (75.00)	8 (100.00)
Vitamin D	11 (91.67)	4 (100.00)	7 (87.50)
Analgesics	2 (16.67)	2 (50.00)	0 (0.00)
Growth hormone	1 (8.33)	0 (0.00)	1 (12.50)
XLH medication adherence, N (%)			
Regularly	10 (83.33)	2 (50.00)	8 (100.00)
Non-consecutive	1 (8.33)	1 (25.00)	0 (0.00)
Rarely	1 (8.33)	1 (25.00)	0 (0.00)
Complications *, N (%)			
Nephrocalcinosis	6 (50.00)	2 (50.00)	4 (50.00)
Digestive disorders (heartburn, diarrhea)	5 (41.7)	2 (50.00)	3 (37.50)
Hyperparathyroidism	1 (8.33)	1 (25.00)	0 (0.00)
History of physical therapy, N (%)	5 (41.67)	4 (100.00)	1 (12.50)
History of surgery, N (%)	5 (41.67)	3 (75.00)	2 (25.00)
History of fractures, N (%)	3 (25.00)	3 (75.00)	0 (0.00)
Deformations of lower limbs, N (%)	12 (100.00)	4 (100.00)	8 (100.00)

SD, standard deviation; XLH, X-linked hypophosphatemia
*Multiple responses were allowed for health clinic services, treatment history, and complications.

Table 2. Topics about burden of XLH

Topic	Self-report of
Clinical symptom	Short stature
	Pain
	Tired/fatigue
	Craniosynostosis
	Sport/exercise
	Dental problems
Emotional burden	Depression
	Frustration
	Guilt
Financial burden	Time loss
	Medical expenses
Treatment	Diagnosis odyssey
	Frequent hospital visits
	Lack of disease expertise
Social impacts	Bullying
	Academic/work difficulties
Others	Impacts on family
	Waking in the night due to pain

Figure 1. Word clouds showing the most frequently used words across open-ended questions related to XLH

- We counted the top 100 words mentioned by patients with XLH during the interviews.
- Symptom burdens: "leg," "dental," "growth," and "height."
- Treatment burdens: "hospital," "treatment," and "diagnosis."
- Social impacts: "gaze," "school," and "work."
- Emotional burden: "psychology," "mental,"

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