

Online Survey to Characterize the Burden of Illness in Children with X-linked Hypophosphatemia (XLH)

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INTRODUCTION

X-linked hypophosphatemia (XLH) is a rare genetic disorder of renal phosphate wasting and defective bone mineralization caused by high circulating levels of fibroblast growth factor 23 (FGF23) that impair normal phosphate reabsorption in the kidney. XLH symptoms present in childhood and include hypophosphatemia, rickets, bowing of the legs, short stature and gait disturbances that do not fully resolve despite standard of care treatment with oral phosphate and vitamin D metabolites. Limited information is available on the impact of the disease on the quality of life of affected children. Therefore, we conducted an international online survey of parents/caregivers of children with XLH to learn more about their disease experience.

OBJECTIVE

To achieve a better understanding of the disease course of XLH, characterize the disease burden, and assess the disease impact on health-related quality of life in children.

METHODS

Parents completed an IRB approved, online questionnaire on behalf of children with XLH in English and French. All participants were required to provide electronic consent before completing the survey. The survey includes questions on the following:

- Demographics
- Current symptoms/conditions
- Parent completed outcome questionnaires to assess pain, disability and quality of life
 - Pediatric Orthopedic Society of North America Patient Outcomes Data Collection Instrument (POSNA PODCI)
 - SF-10 Health Survey for Children (SF-10)
- Diagnostic history
- Current treatments used to manage XLH
- Medical/surgical history
- Use of assistive devices for walking

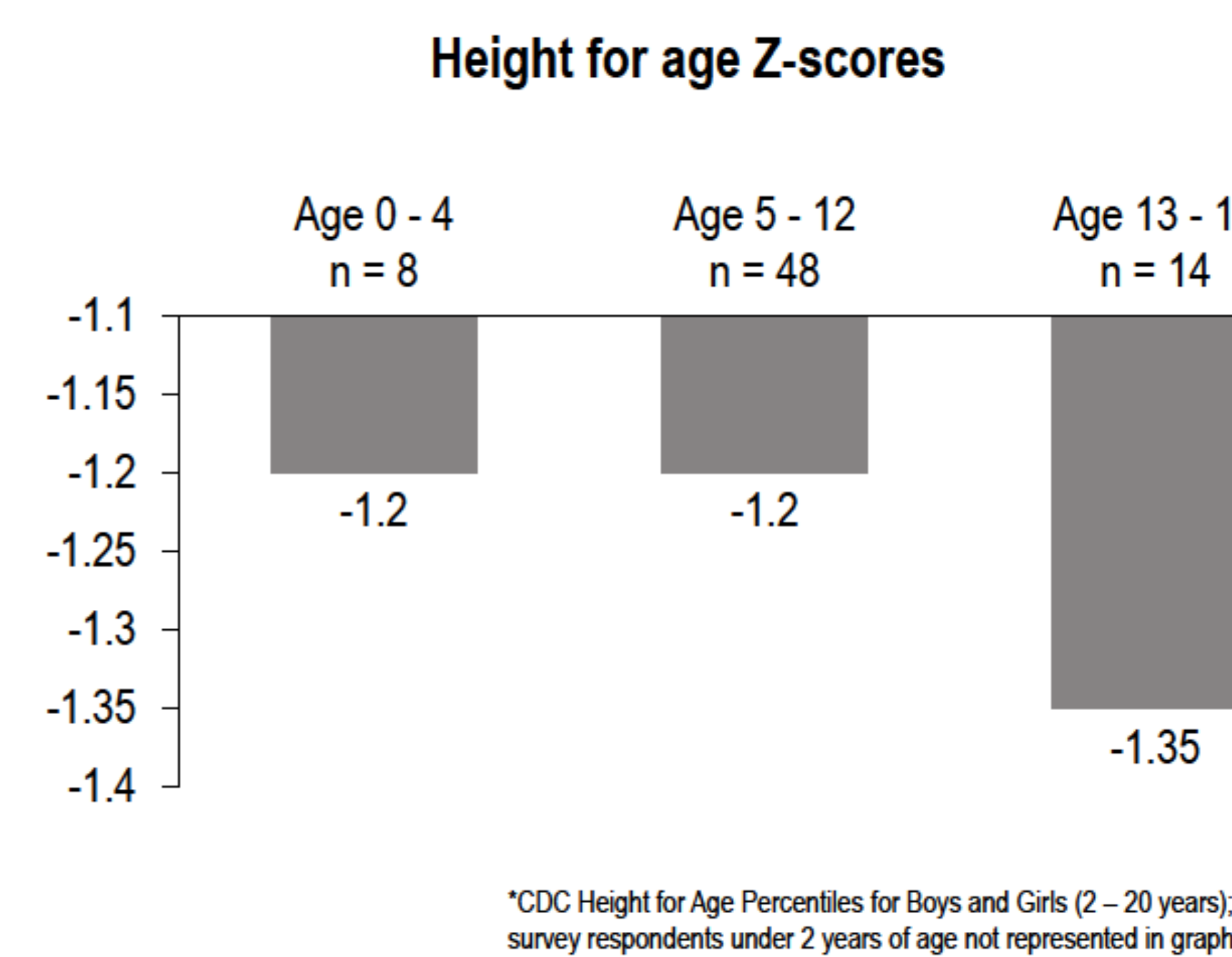
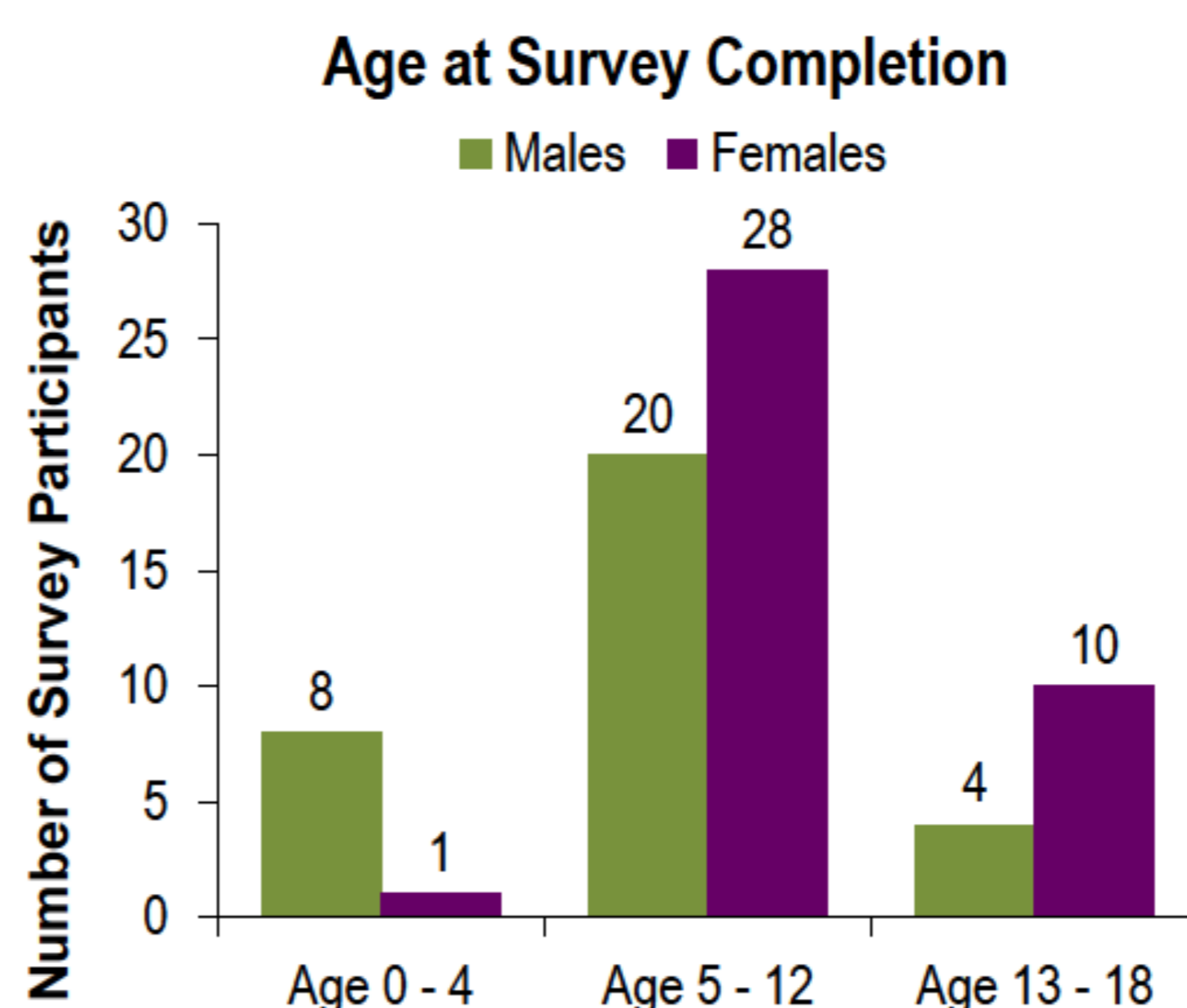
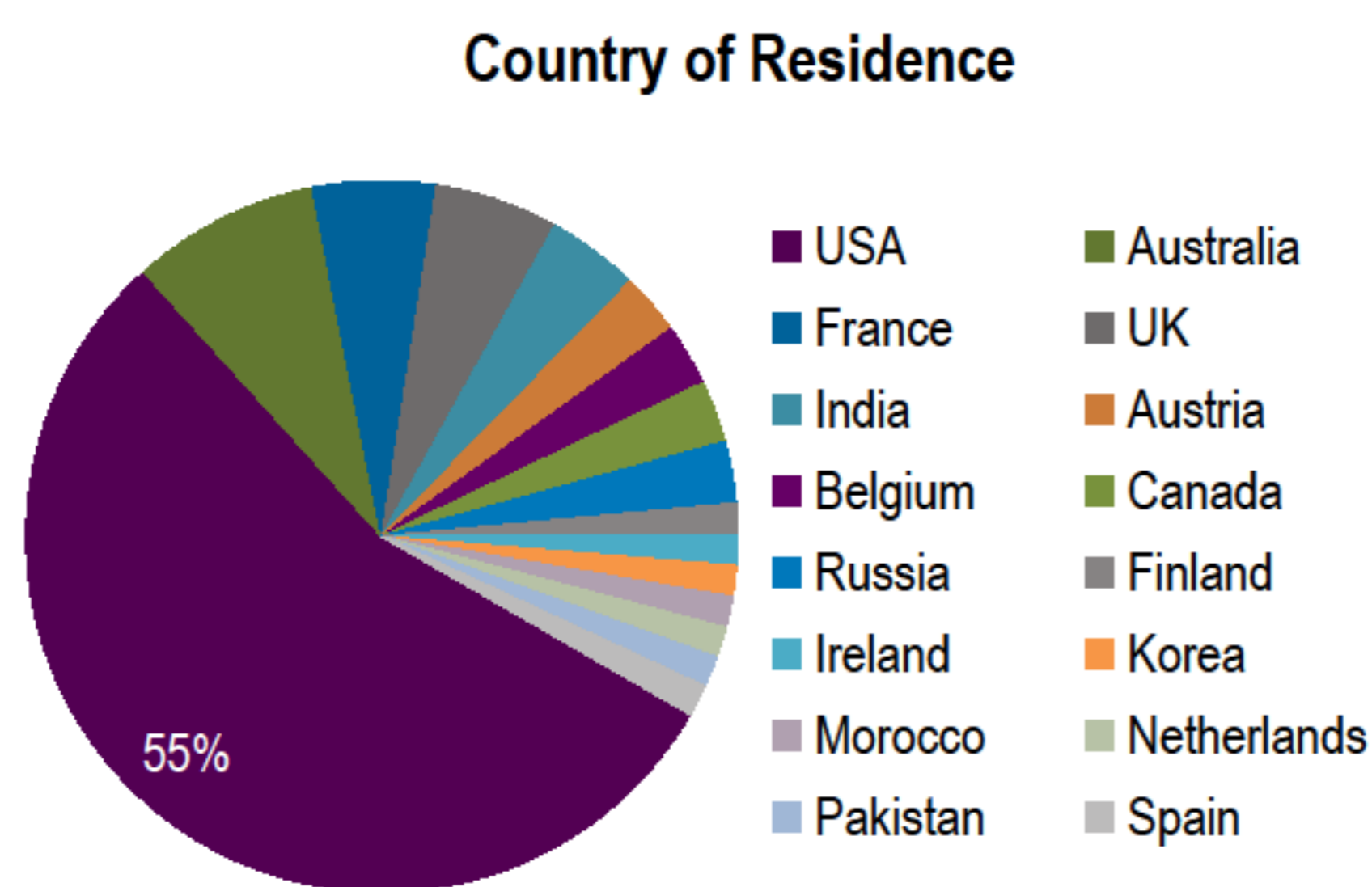
Data collection began on June 20, 2014 and is ongoing

RESULTS

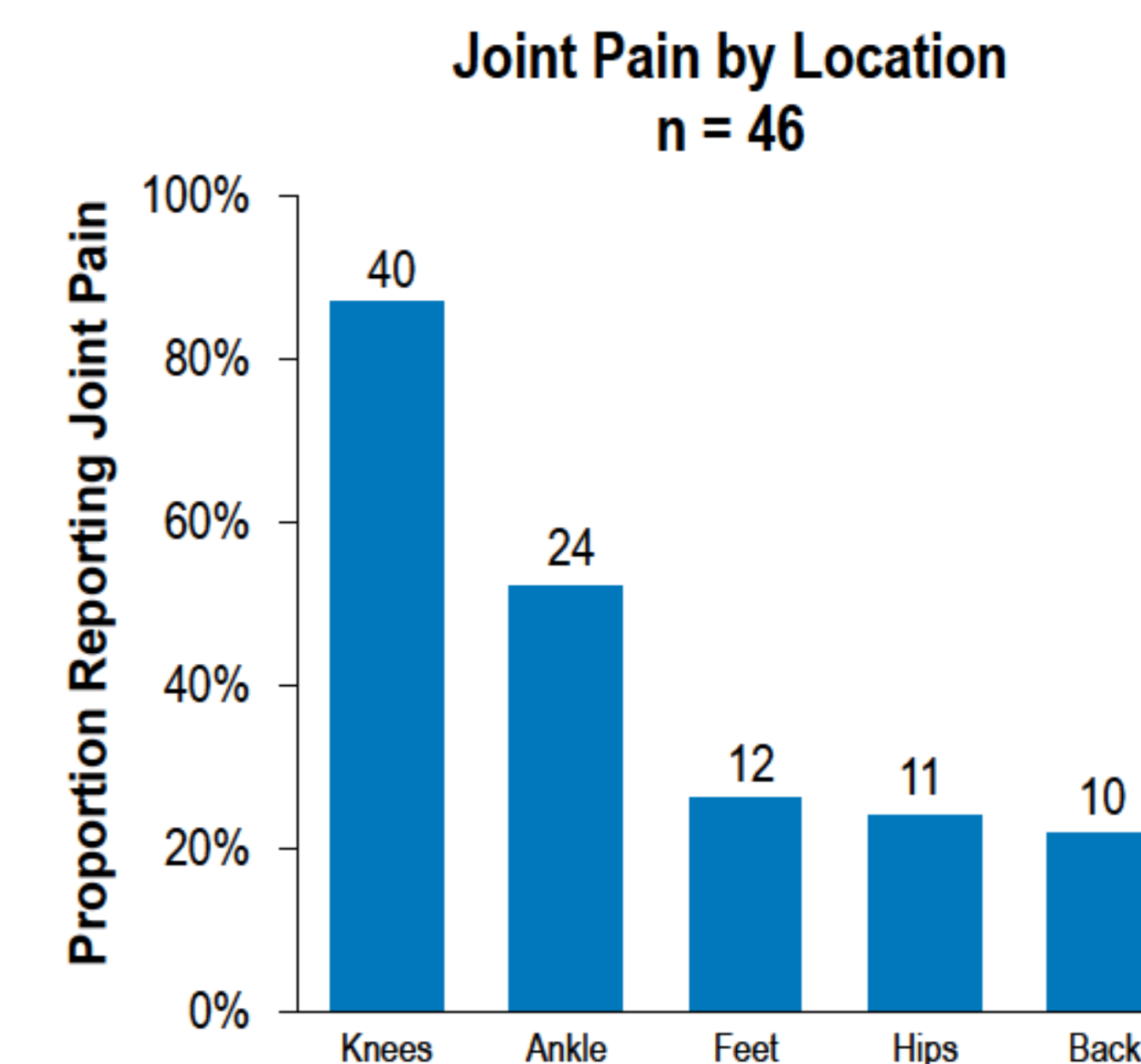
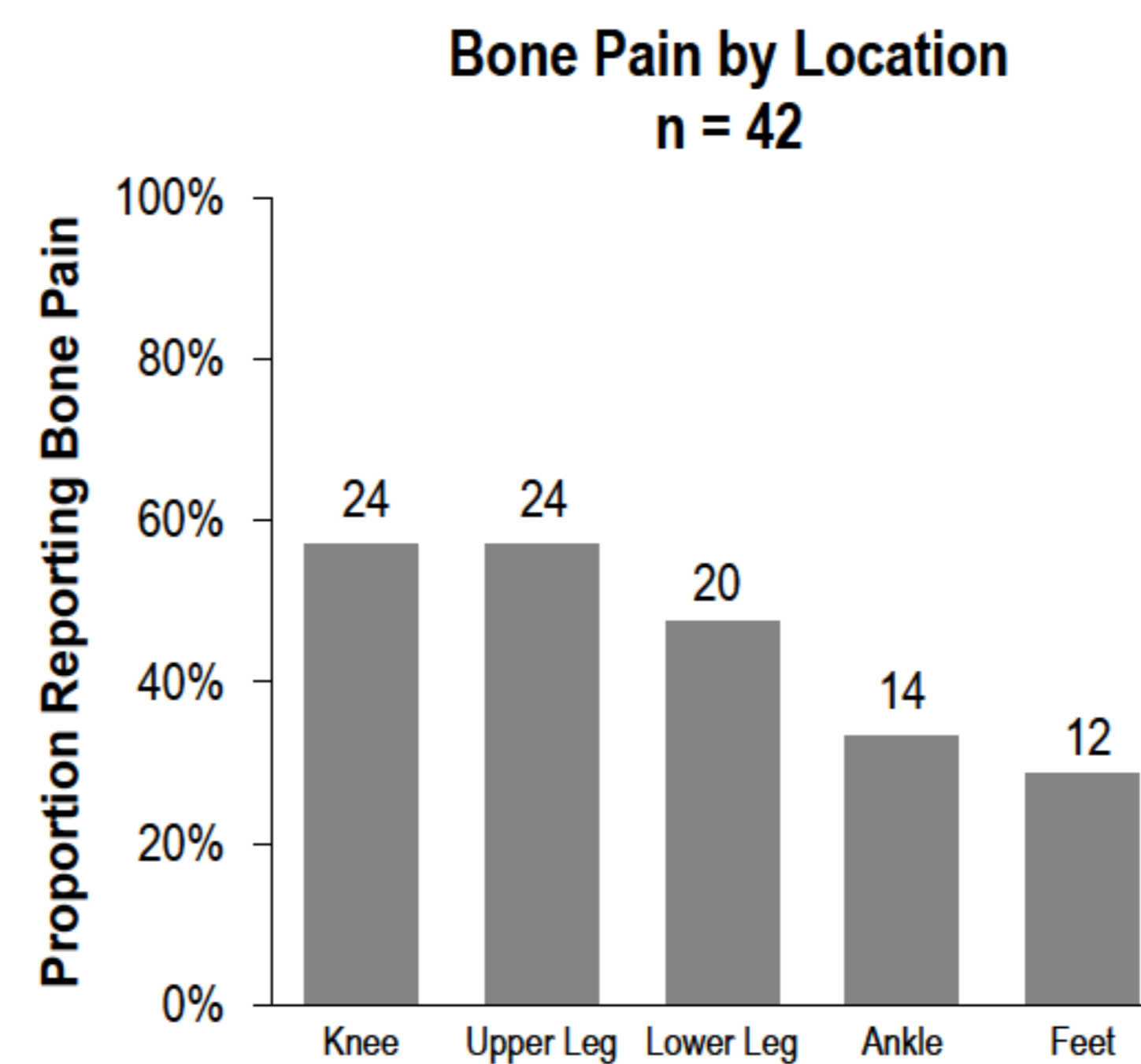
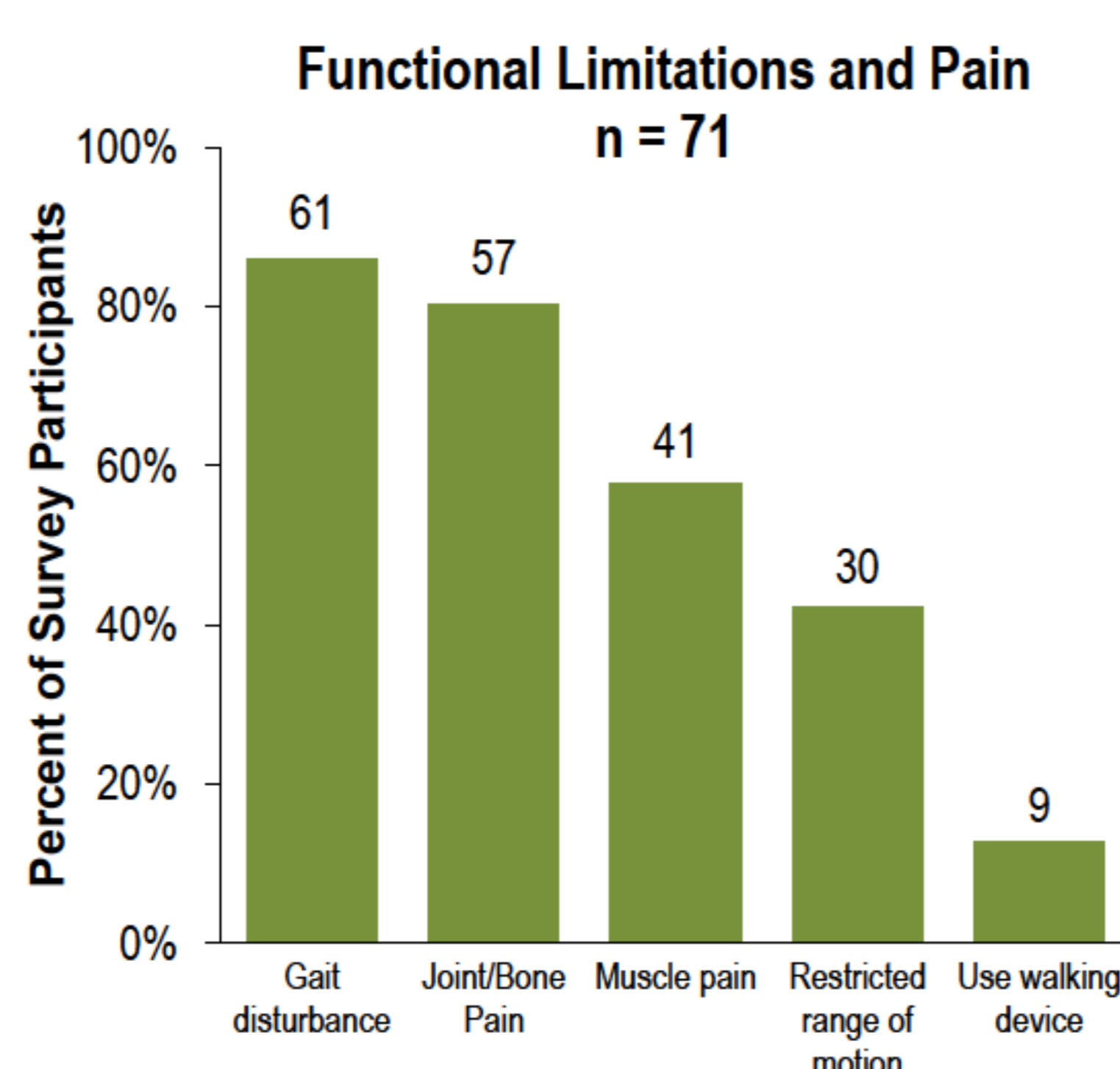
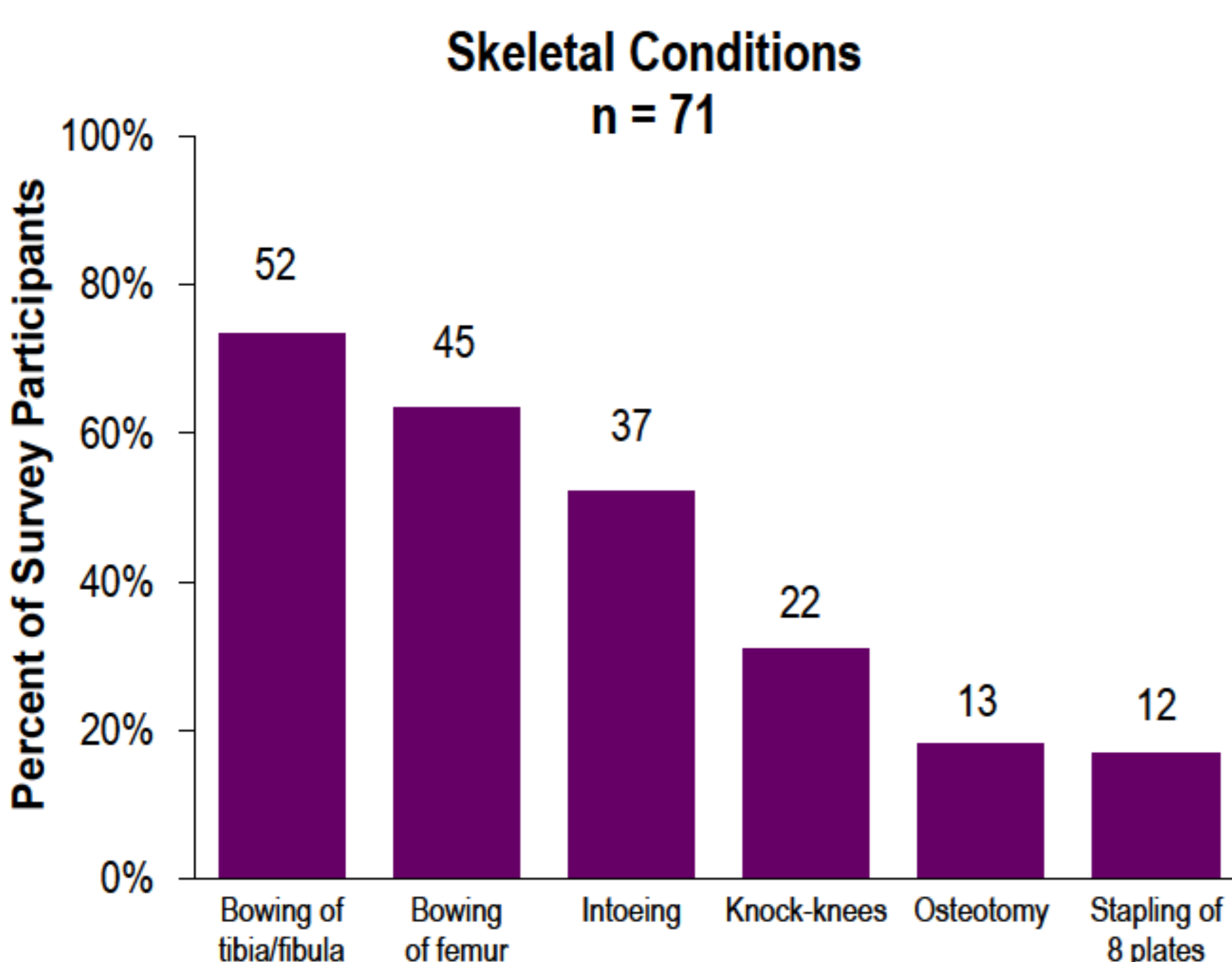
Responses were received for 71 children (39 females; 32 males) from 16 different countries as of April 24, 2015

Age; years	Mean	Median	Range
At survey completion	9.0	9.0	1.0 – 18.0
At first symptoms of XLH	1.4	1.0	0.0 – 12.0
At diagnosis of XLH	2.1	2.0	0.0 – 17.0

Current treatment for XLH	
	%
Phosphate & Vitamin D metabolites	98.6



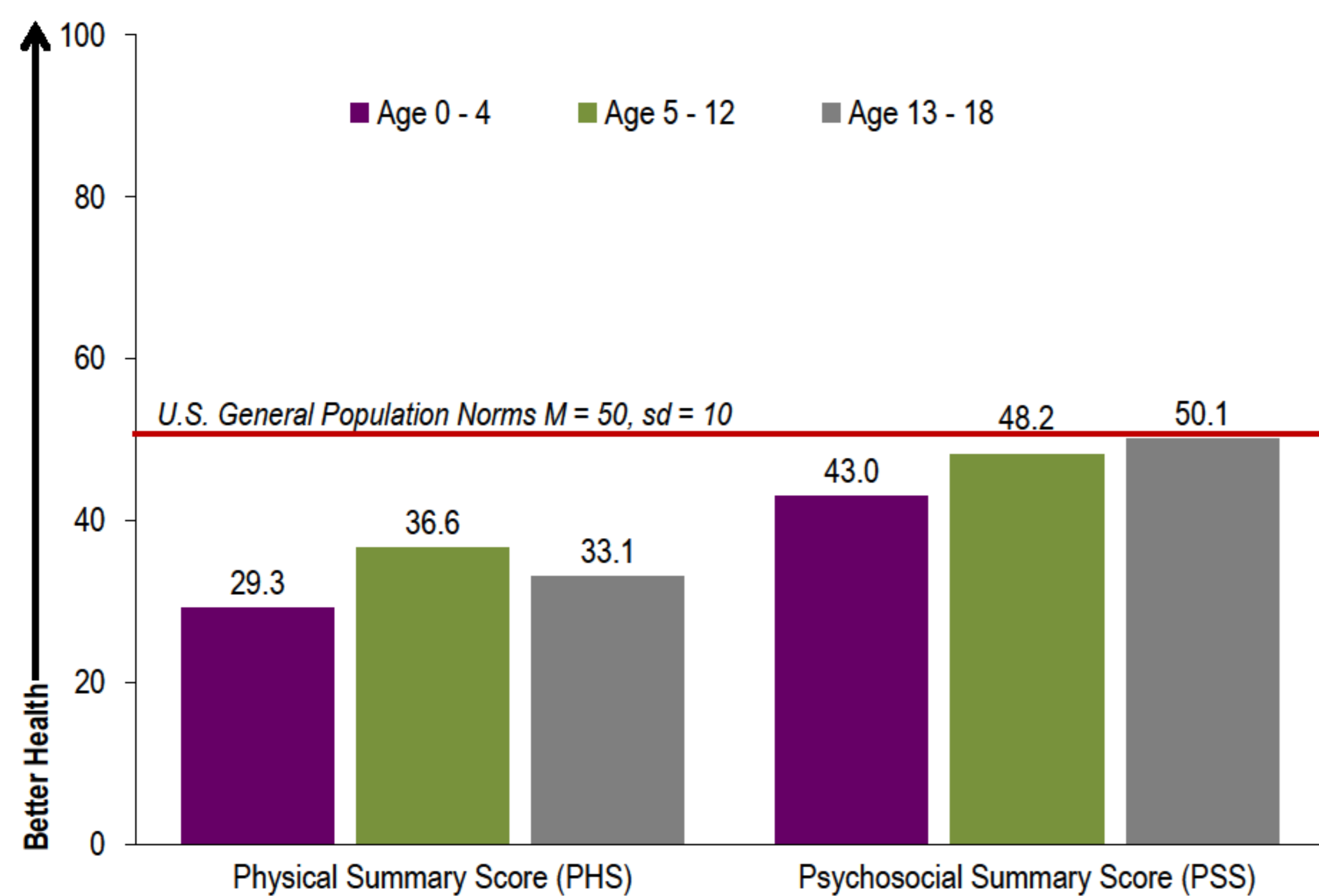
BURDEN OF DISEASE



QUALITY OF LIFE

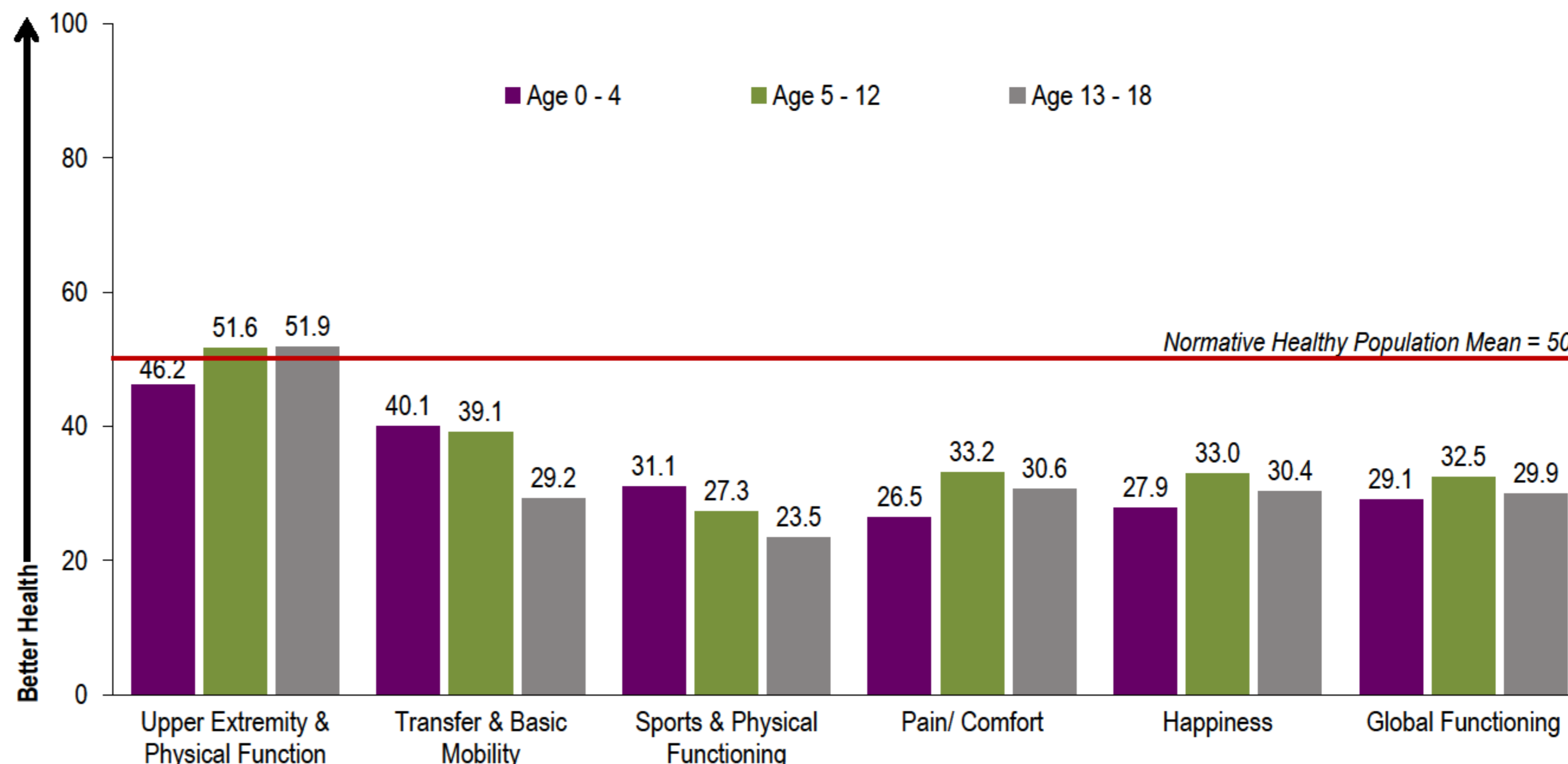
SF-10

- 10-question parent-completed survey
- Evaluates the following concepts: physical functioning, role/social emotional-behavioral, role/social physical, bodily pain, general behavior, mental health and self esteem



POSNA PODCI

- Parent-reported questionnaire to assess overall health, pain and ability to participate in normal daily activities, as well as in more vigorous activities associated with young people
- Six Scales: Upper Extremity and Physical Function, Transfer and Basic Mobility, Sports/Physical Functioning, Pain/Comfort, Happiness, and Global Functioning



CONCLUSIONS

- Significant XLH-related skeletal abnormalities persist despite prolonged treatment with phosphate and vitamin D metabolites.
- Lower extremity bone and joint pain resulting from weight bearing on weak bones and misaligned joints appear to be common in children with XLH.
- Mobility and gross motor function are impacted by the skeletal abnormalities and pain and impair quality of life throughout childhood.
- Alternative treatments options are needed to minimize the burden of disease in children with XLH.

Disclosures: Authors with Ultragenyx Pharmaceutical Inc. affiliation are employees and shareholders of Ultragenyx Pharmaceutical Inc., the study sponsor.

