

# XLH Community Impact Survey

## Data Report from 2024 XLH Community Impact Survey and Interviews

August 2024

# Survey Overview & Respondent Demographics

# Background & Objectives

## Background

- XLH\* is rare, and little is known about its impact on a person throughout life
- Unmet needs of the U.S. XLH community extend beyond the burden of symptoms to accessing care, navigating care transitions and day to day activities, ability to work, etc.; data to illustrate the impact of these needs are lacking
- To address this gap, Kyowa Kirin and The XLH Network joined together to conduct a survey to quantify and deepen understanding of the current experience of adult and child XLHers in the U.S.

## Objectives

- To generate data to support the anecdotal experiences of XLHers in the U.S.
- These research findings may be used to:



### \*About XLH

- X-linked hypophosphatemia (XLH) is a rare, lifelong condition caused by a low level of phosphorus in the blood
- It can affect the bones, muscles, and teeth of both children and adults
- XLH is primarily a hereditary condition and affects up to 1 in 20,000 people

# Research Methods

Primary research was conducted with adult XLHers and caregivers of child XLHers who reside in the United States

## Inclusion Criteria

- Participants must have:
  - Been aged 18+ and diagnosed with XLH or the parent/caregiver of a person diagnosed with XLH
  - Resided in the U.S.

\*If a child was between age 15 and 17 years, they had the option to fill out an assent form and were able to participate in the interview alongside their caregiver

\*If more than 1 person in a family was diagnosed with XLH, they were able to revisit the site and participate again for each diagnosed individual

## Online Survey

- Participants visited a secure Engage Health site to complete the survey
- Survey captured:
  - Consent & demographics
  - Healthcare provider (HCP) & insurance information
  - Access to expert, quality care
  - Psychosocial impact of XLH
  - Financial burden of XLH
- Participants could opt-in to participate in an interview

## Telephone Interview

- Engage Health conducted one-hour interviews to probe further into participants':
  - Background
  - Impact on daily activities
  - Physical symptoms
  - Financial impact
  - Access challenges
- Participants were paid \$100 as compensation for their time

# Survey Limitations and Notes for Interpretation

- Research survey and interviews were fielded from November 2023 – February 2024
- Research was conducted in the U.S. only, and reflects a respondent base with connections to The XLH Network and Kyowa Kirin through XLHLink and/or Kyowa Kirin Cares
- The survey was designed to learn about the lived experiences of XLHers and was not intended to assess statistical significance or determine correlation or causation
- In some instances, the sample sizes are small and may only reflect the perspectives of the participants
- Financial questions ask respondents to reflect on the past year only, and results may not reflect the long-term financial impact of XLH
- Where possible, survey findings are contextualized by insights gathered through follow-up interviews. Note, however, findings throughout may surface additional questions which were not addressed by this research and may be further explored in the future

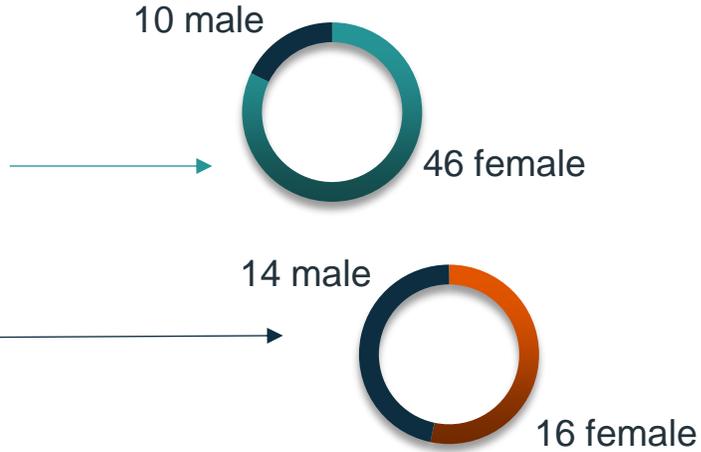
# Study Overview

## 87 Survey Completions

56 adults

29 caregivers\*

2 children



Average age for adult XLHers was 49.4 years and 9.0 years for child XLHers

Average time since diagnosis was 42.2 and 7.0 years for adult and child XLHers, respectively

Insufficient data to discern the number of spontaneous vs. genetic cases

\*Throughout, "child XLHer" includes experiences of children diagnosed with XLH, as reported by a caregiver or in a child / caregiver dyad  
\*Note: there was 1 caregiver who did not provide the sex of the child XLHer

## 56 Qualitative Interviews

36 adults

20 caregivers

## Recruitment channels

The XLH Network | XLHLink | KKCaress

# Geographic representation across the United States



N=87, 56 adult XLHers and 31 pediatric XLHers (including 29 caregivers and 2 child XLHers)

Broad distribution across the US:

69



Urban

14



Rural

When data was considered using the Rural Health Information Hub's distinction of Urban vs. Rural\*:

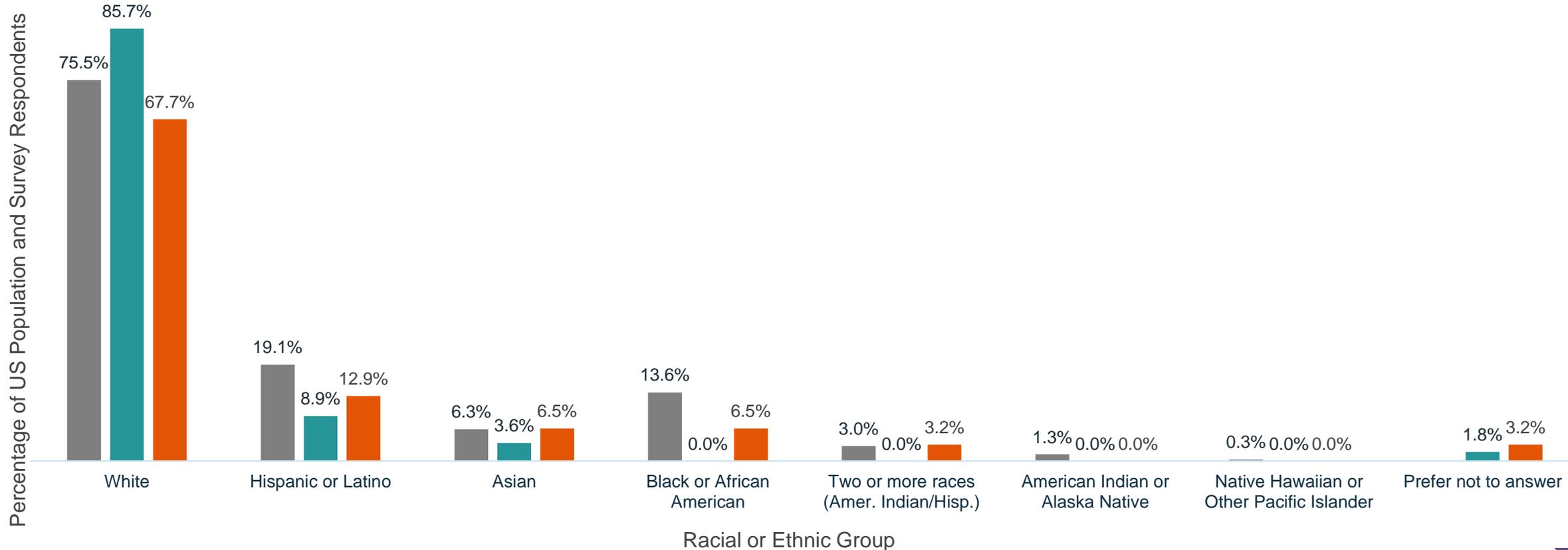
- Most (79.3%, 69) respondents were from an urban setting
- 16.1% (14) were from a rural setting
- 4.6% (4) were undetermined as they did not share location

\*Found at [www.ruralhealthinfo.org](http://www.ruralhealthinfo.org)

# While the majority of XLHers represented in the study were white, there was representation from other racial groups, particularly among child XLHers. Hispanic, Black/African American, American Indian/Alaska Native, and Hawaiian/Pacific Islanders were underrepresented in the study

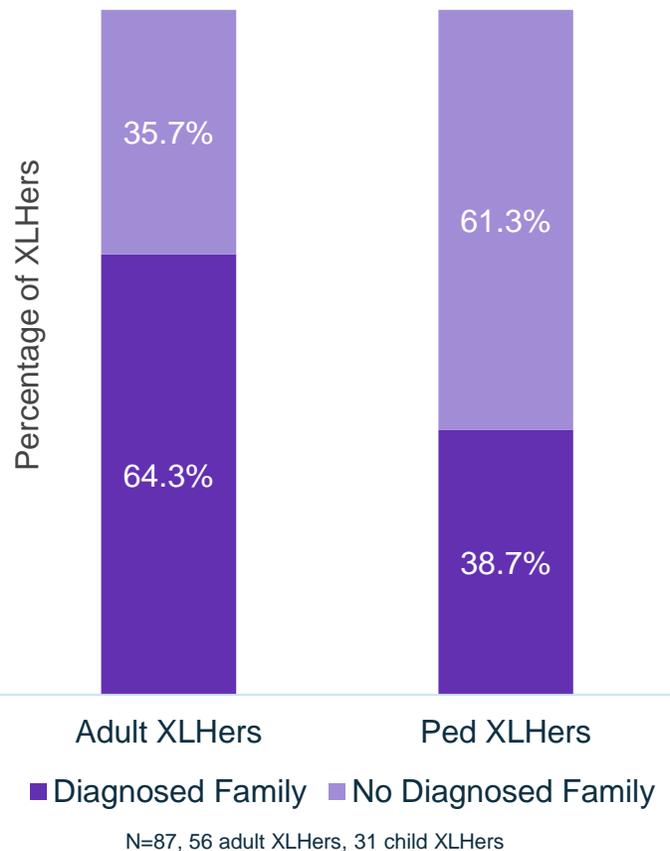
■ US Overall ■ Adult XLHers ■ Child XLHers

**Note:** The US Overall percentages are for comparison with this study and were sourced from the US Census, as of July 2023. Link: <https://www.census.gov/quickfacts/fact/table/US/PST045223>

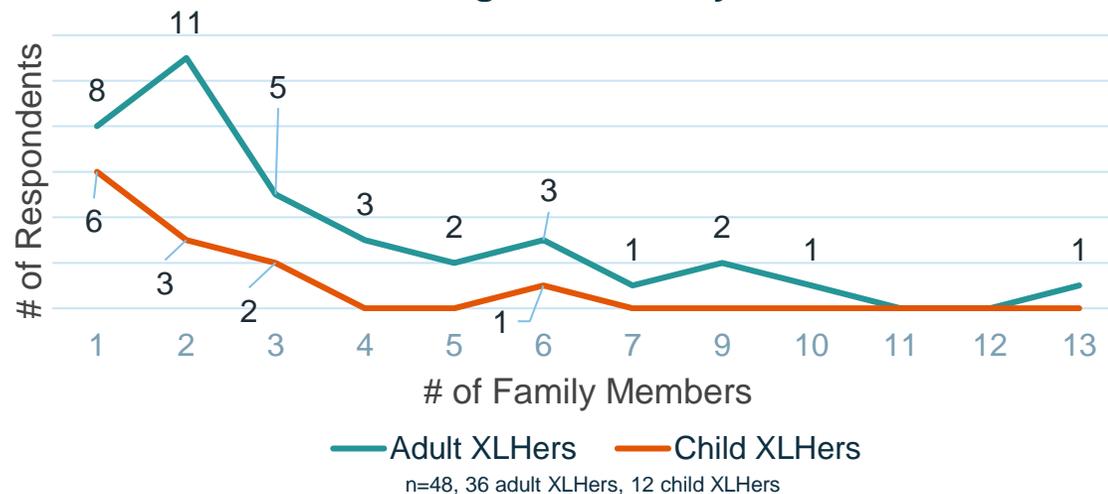


# Adult XLHers were more likely to have diagnosed family members; most had either 2 or 3 family members (most often a mother, daughter, sister, or son)

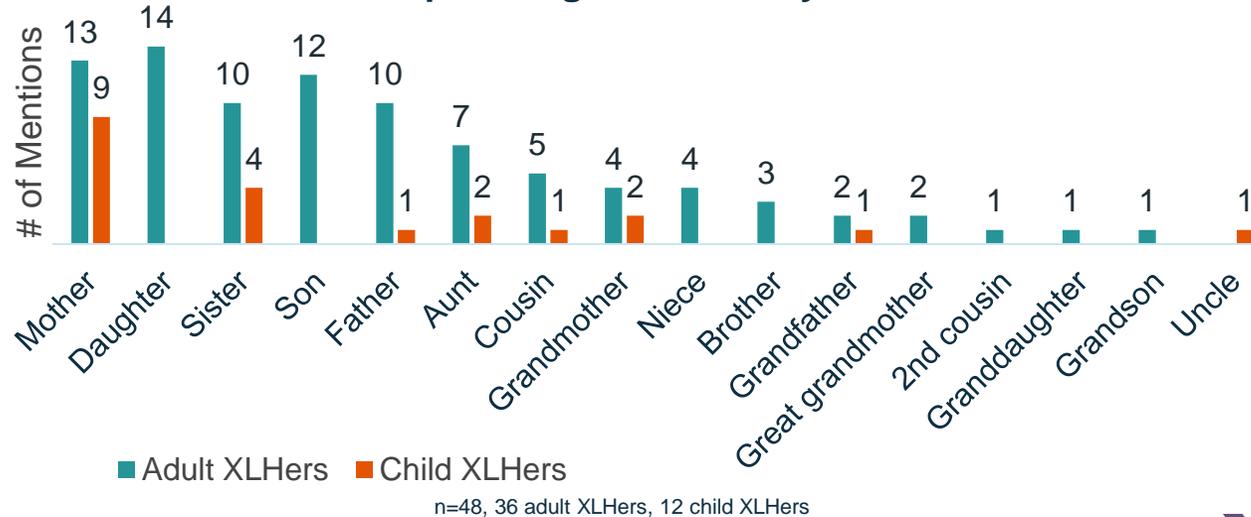
Percentage of XLHers with Diagnosed Family Members



Number of Diagnosed Family Members



Relationship of Diagnosed Family Members



# Executive Summary

## 21% of adult XLHers saw 5+ healthcare providers (HCPs) to reach a diagnosis

Access to Expert  
Quality Care

- Adult and child XLHers saw 2.3 HCPs prior to diagnosis on average, but adults were twice as likely to see 5+ providers
- Bowed legs was the most frequently mentioned initial sign/symptom that led to diagnosis, followed by family history of XLH, and milestone delays (e.g., walking)
- Diagnosis most often made by an endocrinologist for all XLHers
  - Child XLHers also commonly diagnosed by a geneticist
- On average, adult XLHers rated their diagnosing HCP's knowledge of XLH lower than caregivers of child XLHers
  - Adult XLHers recognized medical knowledge "at that time" they were diagnosed was less robust

*“ There were no information gaps. They knew as much as they could at that time, the doctor that diagnosed me is also one of the leading doctors in XLH now.*

*– Adult XLHer, rated diagnosing HCP knowledge as high*

*At the time, they didn't know how to diagnose or treat XLH. I had my first surgery in June in 1974, but beyond that the doctors didn't know much about how to treat it properly.*

*– Adult XLHer, rated diagnosing HCP knowledge as low*”

QX #8: How many physicians did you/your child see for XLH-related issues prior to receiving a diagnosis of XLH? (n=64, 34 adult, 30 child)

QX #39: I want to talk a bit about your "journey" of how you got to the doctor who diagnosed you, as well as the others. Please tell me how you got to the point of being diagnosed with XLH (n=56, 36 adult, 20 child)

QX #9: Please select the option that best describes the specialty of the healthcare provider who diagnosed you/your child with XLH: (N=87, 56 adult, 31 child)

QX #13: On a scale of 1-10, with 1 being extremely poor, and 10 being excellent, how would you rate the expertise of each HCP related to their knowledge of XLH? (N=87, 56 adult, 31 child)

**Kyowa KIRIN**

**XLH**  
NETWORK

# XLHers are generally satisfied with their current healthcare provider (HCP) but have had challenging care experiences

Access to Expert  
Quality Care

- Barriers to care cited include travel distance to providers, long waits at providers, lack of HCP knowledge, and fear of moving to a new HCP
- Majority (79%) reported their main XLH HCP is an endocrinologist
- XLHers travel nearly 90 miles, on average, one way to access their current primary XLH medical provider
- Perceived knowledge of primary XLH HCP rated fairly high, and somewhat higher among child vs. adult XLHers
- Positive care experiences driven by HCP involvement in XLH research, XLH training, and patient familiarity

“ Any time I tried to get care it's been difficult especially with endocrinology. When I lived in a small town, and I had to travel hours for even a little bit of knowledge. When we moved to a new state, I tried to find one in my city and couldn't get them to take my case. That was really frustrating, and I had to travel 75 miles each way to the doctor who hadn't ever heard of XLH.

--Adult XLHer

QX #60: In the survey you also rated your experience with various items impacting access to care. Can you tell me more about this? (n=56, 36 adult, 20 child)

QX #10: Please select the option that best describes the specialty of the healthcare provider who is the main healthcare provider related to XLH for you/your child: (N=87, 56 adult, 31 child)

QX #14 and 15: On a scale of 1-10, with 1 being extremely poor, and 10 being excellent, how would you rate the process of scheduling a visit and the travel distance with the main healthcare provider for XLH: (N=87, 56 adult, 31 child)

QX #13: On a scale of 1-10, with 1 being extremely poor, and 10 being excellent, how would you rate the expertise of each HCP related to their knowledge of XLH? (N=87, 56 adult, 31 child)

# Respondents rated the XLH knowledge of dental providers as quite low, though most reported their dentist was willing to learn about XLH

Access to Expert  
Quality Care

- Nearly 8 in 10 XLHers reported currently receiving dental care at the time of the study, with children slightly more likely to have care
  - 86% of XLHers with private insurance had dental care
  - 67% of XLHers with government insurance had dental care
- XLHers rated their dental providers' knowledge of XLH at around 5 on a 10-point scale
- 75% of XLHers with a current, consistent dentist said provider was receptive to learning about XLH
- XLHers travel 12.6 miles / 0.6 hours one-way to current dentist on average

*The dentist didn't know about XLH when we first went, but the more that we go over the years, she is more understanding of what is going on with my child's teeth, the decay issues and soft teeth, and she relates it back to XLH.*

*-- Caregiver of child XLHer*

QX #11: Please select all options that describe the specialty of the healthcare providers who you/your child currently see for various issues related to XLH. (N=87, 56 adult, 31 child)

QX #13: On a scale of 1-10, with 1 being extremely poor, and 10 being excellent, how would you rate the expertise of each HCP related to their knowledge of XLH? (N=87, 56 adult, 31 child)

QX #37: For your dental care, do you have a primary dentist? Did you find them on your own vs. referral? (n=56, 36 adult, 20 child)

QX #14 and #15: n a scale of 1-10, with 1 being extremely poor, and 10 being excellent, how would you rate process of scheduling and the travel distance to see the dental care provider: (N=87, 56 adult, 31 child)

# Most adult XLHers interviewed (94%) did not formally transition care from pediatric to adult healthcare provider (HCP)

Access to Expert  
Quality Care

- Top cited reasons for lack of care transition included:
  - Patients told “disease would go away” after growth stopped
  - Difficulty finding new HCP
  - XLHer did not follow-up with care
- Perception of transition was highly polarized, with two in three XLHers rating the difficulty of the experience at 1 or 10

“ *I wish they would’ve had packets of information for those aging out and transitioning to adult care... I didn't really learn enough to know... it would have been a big help to not let things keep going downhill.*

– Adult XLHer

*I aged out [of pediatric care] when I became an adult and I didn't have any other doctors I could go see. I didn't have anywhere to turn... and they didn't help me find a new person. There was a long gap and the next 10 years was sporadic care here and there until maybe 2016 when I found information online.*

– Adult XLHer

QX #30: At the time you were transitioning from pediatric to adult care, how difficult was it to find a new doctor on a scale of 1-10 with 1 being not difficult at all and 10 being extremely difficult? (n=56 adult)

QX #63: Tell me more about transitioning from pediatric to adult care...what could have gone better or been done to improve the transition? (n=36 adult)

# Adult XLHers reported significant physical impact from XLH, including substantial pain and dental, hearing, and sleep issues. Child XLHers experienced these issues, but at a lower rate

- XLHers reported the most burdensome physical symptom was pain, followed by lack of flexibility/stiffness (for adults) and physical appearance (for child), then lack of energy regardless of age
- Most adult XLHers reported at some point having dental cavities (97%), dental abscesses (87%), early tooth loss (84%), and no/poor enamel (74%)
  - Half of child XLHers had cavities and 40% had no or poor enamel
- Most adult XLHers reported hearing issues, including tinnitus (80%) and partial hearing loss (65%); only 5% of child XLHers had these
- Adult XLHers reported sleep issues more frequently than caregivers of child XLHers, including difficulty falling and staying asleep
- XLHers with high pain and/or sleep burdens rated psychosocial burdens higher than those who did not have these high physical burdens

*“ For as long as I can remember I have dealt with pain. It stands out more when I don't have pain rather than when I do, it's just a chronic thing that affects different areas. You get used to it, but **it's always there**. I am cautious with my pain too. It always concerns me, I am wondering what the pain is from, if is it bone pain or a calcium deposit... wondering what it is and when to go to the doctor. It can be debilitating.*

– Adult XLHer

QX #53: Please rank the 3 most burdensome physical symptoms related to XLH that affect you as a parent/caregiver on a day-to-day basis, in order of importance (n=56, 36 adult, 20 child)

QX #48: Do you/your child experience the following oral/dental health issues and how would you describe the issue: (n=56, 36 adult, 20 child)

QX #49: Do you/your child experience the following hearing health and how would you describe the issue: (n=56, 36 adult, 20 child)

QX #52: Do you/your child experience the following sleep issues and how would you describe the issue: (n=56, 36 adult, 20 child)

# XLHers reported substantial negative impacts of XLH on physical and mental health, resulting in difficulties with activities of daily living, absenteeism, and reduced productivity at work or school

- Pain was the most burdensome issue reported, followed by mental health and ability to stay asleep
  - Absenteeism, reduced productivity at work or school, and challenges with relationships were reported as somewhat burdensome
- XLHers reported substantial mental health concerns, e.g., anxiety and fear
  - Adult XLHers noted high prevalence of depression, social isolation
  - Child XLHers experienced self-esteem issues stemming from physical appearance and XLH impact on social participation
  - Few XLHers sought help from mental health professionals (13% adult, 3% child)
- Adults experienced greater impact of XLH on activities of daily living than children; most common challenges were getting dressed and housework
- Adults rated medical trauma nearly twice as burdensome as children; most common cause was surgery recovery for adults, needle phobia for children

“ Across my lifespan, the impact has varied. As a kid and through high school, it was about **looking different**, not hearing as well, and awkward “peer stuff”. As I got older (in my 20s), everyone was getting married, and I realized it was going to be different for me...as I'm getting older it's more challenging as my **mobility and restrictions** have increased. The hearing issues have the biggest social impact...I can't join groups or meetups because it's hard to hear.

– Adult XLHer ”

QX #16: On a scale of 1-10, with 1 being "no impact" and 10 being "extreme impact", please rate the impact that XLH has had on you/the person diagnosed with XLH in the last year (N=87, 56 adult, 31 child)

QX #51: Do you/your child experience the following mental health issues and how would you describe the issue (n=56, 36 adult, 20 child)

QX #41 On the survey we asked about areas of life impacted by XLH. Tell me more about that (n=56, 36 adult, 20 child)

QX #25 and #11: In the last year, have you paid any of the healthcare costs associated with XLH out of pocket? Please estimate the amount that was spent (on mental health care). Describe what is was spent on (N=87, 56 adult, 31 child)

QX #46: How much do these issues or other XLH-related burdens impact YOUR / YOUR CHILD'S ability to care for YOURSELF/ HIM OR HERSELF / your family; scale of 1-10. Please explain (n=56, 36 adult, 20 child)

QX #44: Of all the psychosocial issues we have talked about, please rank the 3 most burdensome for you/your child, in order of importance (n=56, 36 adult, 20 child)

# The financial burden of living with XLH can be very high; average out-of-pocket costs in the past year were highest for home modifications for accessibility (nearly \$7,000), dental care (\$2,775), and assistive devices/medical equipment (>\$2,000); indirect costs include missing work

## Financial Burden

- Significant variation reported in financial burden of XLH: average percentage of annual household income directed to out-of-pocket costs in the last year was 6% but ranged from 0-40%
- Respondents used a range of financial assistance programs
  - 56% used charitable programs
  - 43% used pharma-sponsored programs
  - 18% used public or private disability insurance
- Most commonly cited indirect financial impacts of XLH were missing work and having to use PTO or medical leave

*I tend to use PTO or work late to make up time. I am lucky to have paid time off even though it is not ideal to use it for visits and not vacation. I also keep a lot of supplies, like I always have lidocaine patches, ibuprofen, supplements, compression gloves, deep tissue massages... that all adds to at least \$1000, probably closer to two [thousand].*

– Adult XLHer

QX #25: In the last year, have you paid XLH-related health costs for mental health care, dental care, physical therapy, pain management, or home modifications? Estimate amount and describe what it was spent on. (N=87, 56 adult, 31 child)

QX #26: In the last year, please estimate the percent of your total annual household income that went to paying out of pocket costs associate with XLH (N=87, 56 adult, 31 child)

QX #20: Do you/your child currently utilize any of the following to help pay for their XLH related needs (e.g. medication/care/other)? (N=87, 56 adult, 31 child)

QX #59: Are there any other ways that XLH has impacted you/your family financially? If so, please describe them (n=56, 36 adult, 20 child)

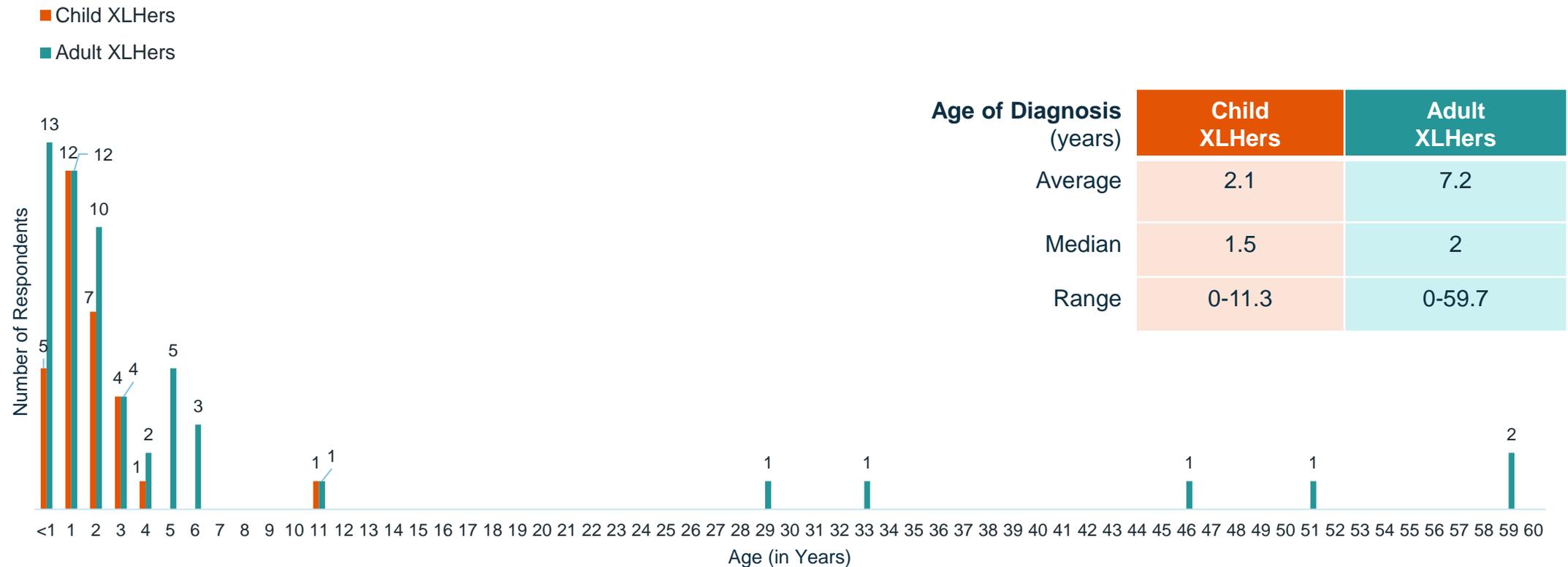
# Detailed Findings

# Access to Expert Quality Care

- XLH Diagnosis
- Main XLH Care Experiences
- Dental Care Experiences
- Care Transition Experiences

# XLHers ranged from 0 - 59.7 years at the time of diagnosis; On average, child XLHers were diagnosed 5.1 years younger than adults

## XLHer Age at Diagnosis



# Diagnostic delays for child XLHers were not as long as those experienced by adults

## Delayed Diagnoses

- Adults with the most delayed diagnoses reported either not having the symptoms “put together” as a complete picture or were misdiagnosed with rickets
  - The 3 adults with the longest delays were diagnosed because their children were tested for XLH
- The longest delays among child XLHers were due to physicians believing the child would grow out of the issue and the time it took to be referred for genetic testing

“*Doctors thought I had a genetically mild case of rickets, where my families have it severely. My short stature wasn't an indicator and my other family members were misdiagnosed with rickets also. **When we tried to identify what was wrong with my son, they eventually led us to do genetic testing.***

– Adult XLHer, diagnosed age 51

*She was a late walker and had a bit different of a walk – at around 2 years old the pediatrician thought it could be **hip development issues** and that she would grow out of it. At 3 she was still ‘waddling’, and we took her to pediatrician again. We were then referred to a physical therapist, then an orthopedic surgeon who thought from x-rays it was some form of rickets. They referred us to endocrinology and confirmed XLH through genetic testing.*

– Caregiver of Child XLHer, diagnosed age 3”

# Family history and a focused caregiver or healthcare provider (HCP) contributed to quicker diagnoses

## Quicker Diagnoses

- Adult XLHers with quicker diagnoses typically either had family already diagnosed or a parent or healthcare provider dedicated to figuring it out
- Quicker diagnoses among child XLHers were made because of known family history or phosphorus levels in blood work prompting genetic testing

“

*I started having symptoms with bowing legs - my mom knew to look because of our family history and took me to get tested, phosphorus was low, confirmed it through x-rays.*

– Adult XLHer, diagnosed age 6 months, currently 40 years

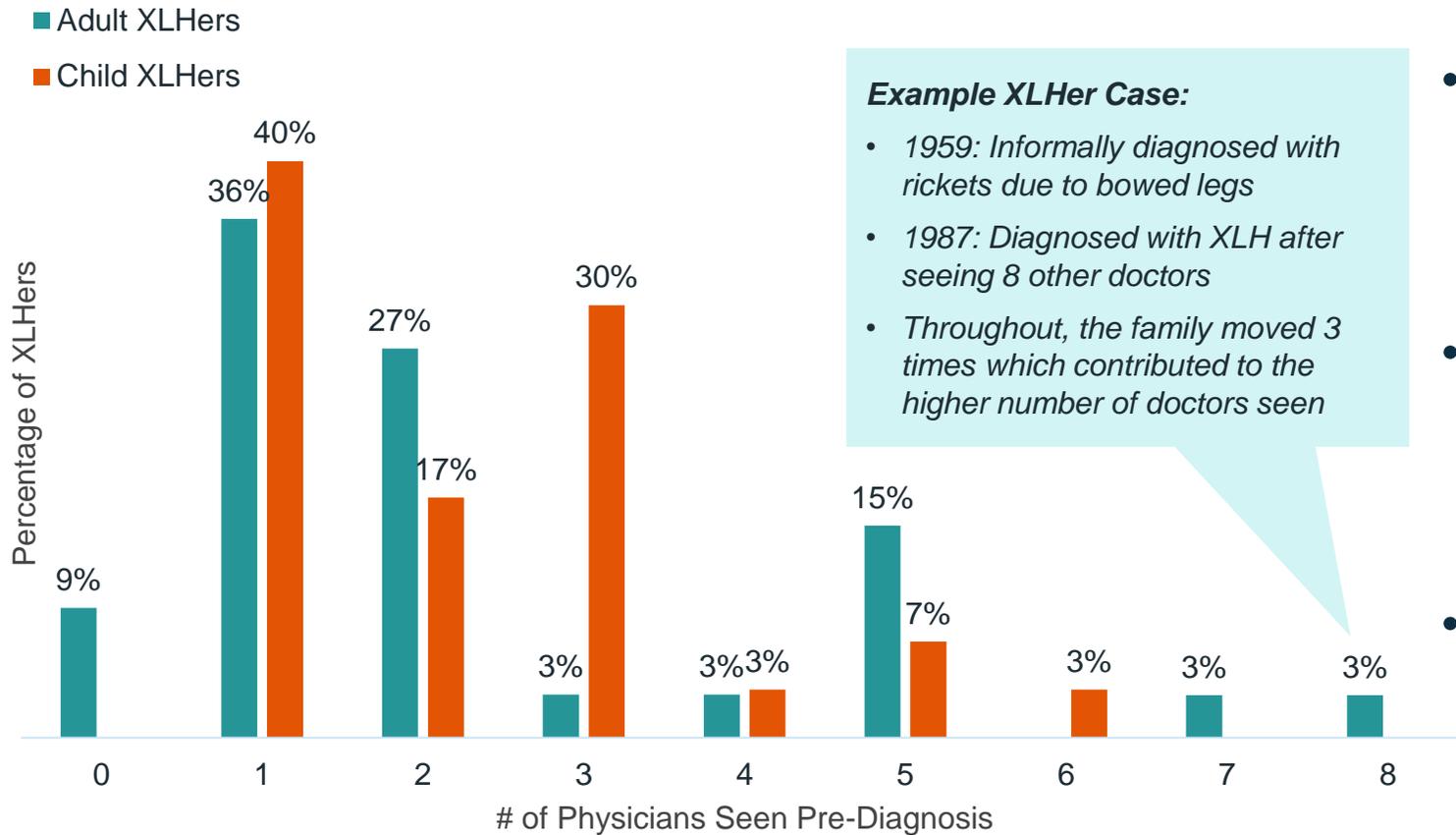
*My child had flaring ribs and his father had febrile seizures so I wanted to avoid those and took him to the ER. They did blood work which showed the phosphorus was off, so they did genetic testing.*

– Caregiver of Child XLHer, diagnosed at 1 year

”

# Though there were XLHers in both groups who saw 5+ HCPs, most were seen by 1-3 HCPs prior to diagnosis

## # HCPs Seen for XLH-Related Issues Pre-Diagnosis\*



**Example XLHer Case:**

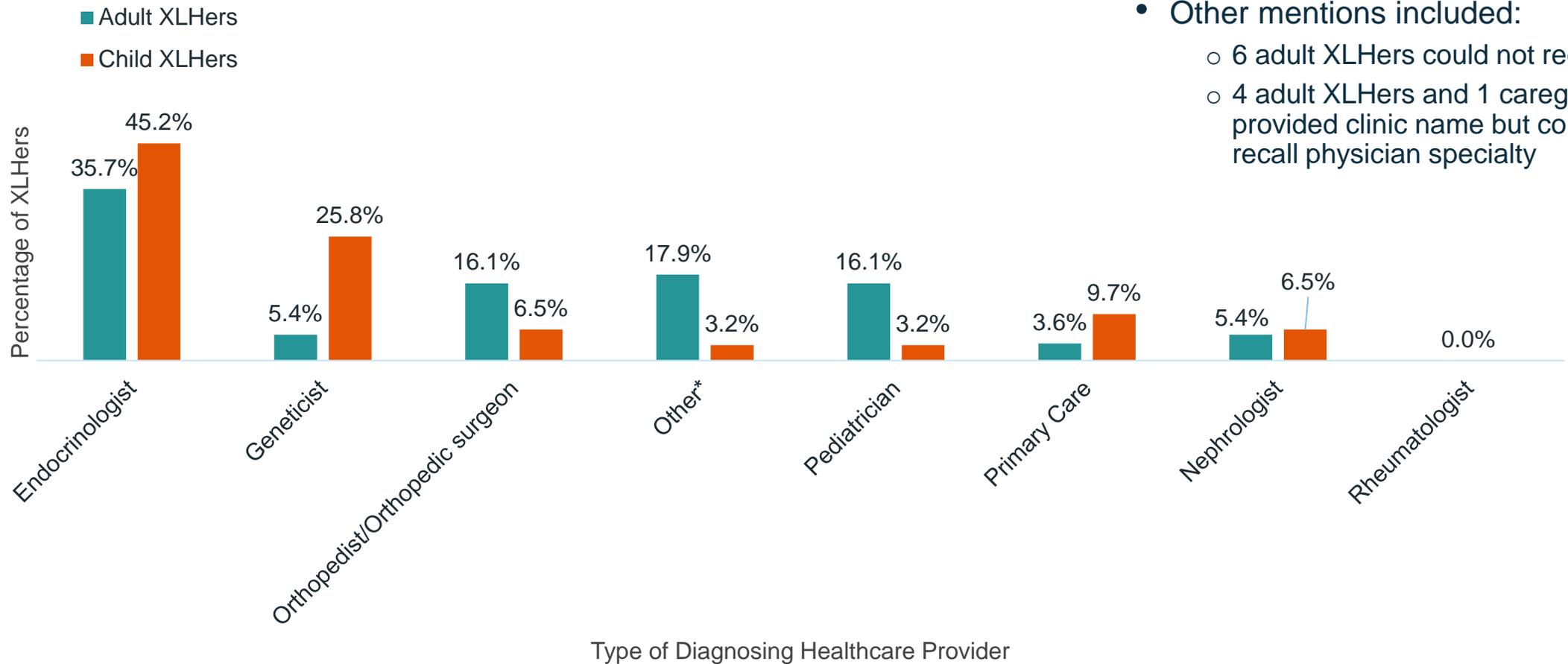
- 1959: Informally diagnosed with rickets due to bowed legs
- 1987: Diagnosed with XLH after seeing 8 other doctors
- Throughout, the family moved 3 times which contributed to the higher number of doctors seen

- Adult and child XLHers both saw on **average 2.3 HCPs** to reach diagnosis
- Child XLHers infrequently saw >3 doctors to reach a diagnosis (likely due to increased availability of genetic testing)
  - The 2 who saw 4+ HCPs were initially misdiagnosed with other conditions
- Adult XLHers seen by 4+ HCPs prior to diagnosis were all diagnosed before availability of genetic testing; their symptoms were either dismissed or not connected to a larger issue
- Note: 3 (9%) indicated 0 in survey responses:
  - Additional detail not available but appears they were diagnosed quickly
  - 1 diagnosed by pediatrician
  - 2 diagnosed by endocrinologist

n=66, 33 adult XLHers, 30 child XLHers; \*Note: 23 adult XLHers and 1 caregiver of a child XLHer did not provide a number  
 QX #8: How many physicians did you/your child see for XLH-related issues prior to receiving a diagnosis of XLH?  
 QX #36: Can you first tell me what types of doctors you saw when trying to get a diagnosis and why you moved from one to the next?

# XLH diagnosis was typically made by an endocrinologist, and for child XLHers, geneticists were also common

## Specialty of HCP Who Diagnosed XLH

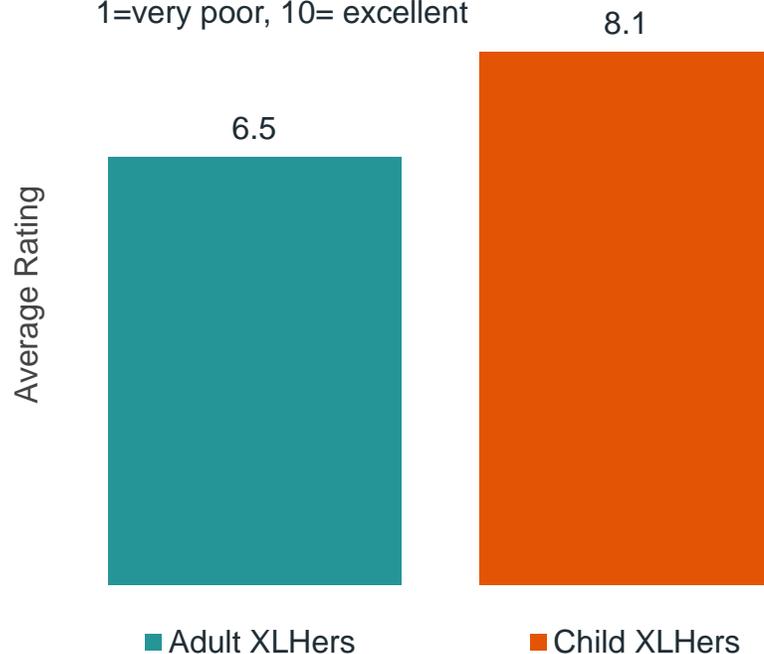


- Other mentions included:
  - 6 adult XLHers could not recall
  - 4 adult XLHers and 1 caregiver provided clinic name but could not recall physician specialty

# On average, adult XLHers rated the XLH knowledge of their diagnosing HCP lower than caregivers of child XLHers; adult XLHers recognized that medical knowledge “at that time” was less robust

## Average Rating of Diagnosing HCP XLH Knowledge

Rating on a scale of 1-10,  
1=very poor, 10= excellent



**“** *Rated diagnosing HCP low (1 out of 10)*

*The doctor didn't know enough about the condition at the time – I just knew I had what was called vitamin D-resistant rickets.*

*–Adult XLHer*

*Rated diagnosing HCP high (10 out of 10)*

*There were no information gaps. They knew as much as they could at that time, the doctor that diagnosed me is also one of the leading doctors in XLH now.*

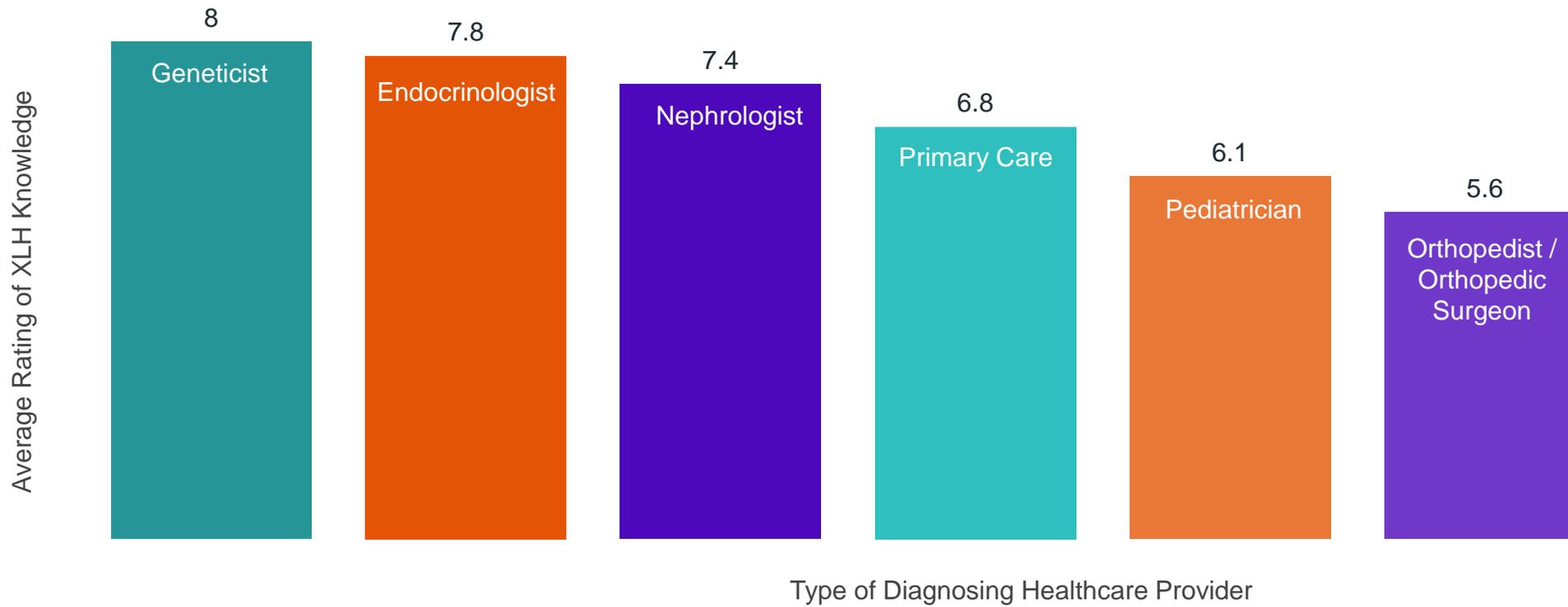
*–Adult XLHer*

**”**

# Across the various diagnosing specialties, XLHers rated the knowledge of endocrinologists and geneticists highest

## Average Rating of Diagnosing HCP XLH Knowledge, by Specialty

Rating on a scale of 1-10,  
1=very poor, 10= excellent



# During interviews, respondents reported bowed legs were the most frequent initial symptom and 20 noted they were diagnosed due to family history of XLH

## Initial Symptoms (# mentions)

- Bowed legs (22)
- Milestone delays: (13)
  - Walking (7)
  - Slow growth (3)
  - Delay in crawling (2)
  - Not rolling over (1 mention)
- Abnormal gait (2)
- Paid attention to gait due to family history (2)
- Knock-knee (2 mentions)
- 1 mention each:
  - In-toeing
  - Falling\*
  - Curvature in stature
  - Abnormal head shape
  - Legs not straightening
  - Flaring ribs
  - Crying a lot as a baby

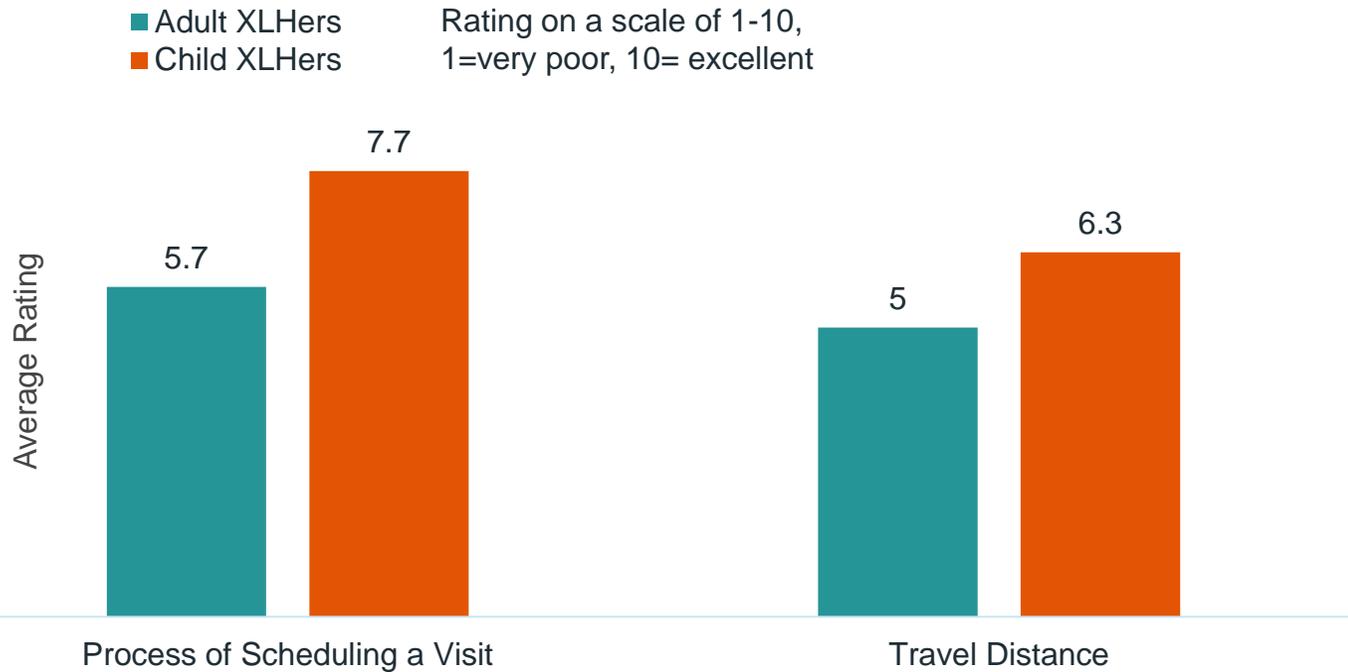
## Other Notes Surrounding Diagnosis (# mentions)

- Patient was subsequently diagnosed because of family history (20)
  - In four cases, an adult patient was diagnosed because their child was diagnosed (4)
- Initial x-rays were conducted (13)
- Abnormal blood work led to further testing (13)
- HCP told parent that child “will grow out of it” (11)
- Specific mention of spontaneous case (2)
- Differential diagnoses included:
  - Blount’s (3)
  - 1 mention each: Fanconi’s, Dwarfism, rare form of diabetes

\*Note: there is insufficient data to assess the impact of pain and stiffness on balance and falls

# Participants traveled on average ~137 miles and 2.4 hours one way to reach the diagnosing HCP

## Average Rating of Scheduling & Travel to Diagnosing HCP



## Distance and Travel Time to Diagnosing HCP

	Adult XLHers	Child XLHers
<b>One-Way Travel Distance (miles)</b>		
Average	152.9	108.3
Range	0-850	0-653
<b>One-Way Travel Time (hours)</b>		
Average	2.6	2.0
Range	0-22	0-10

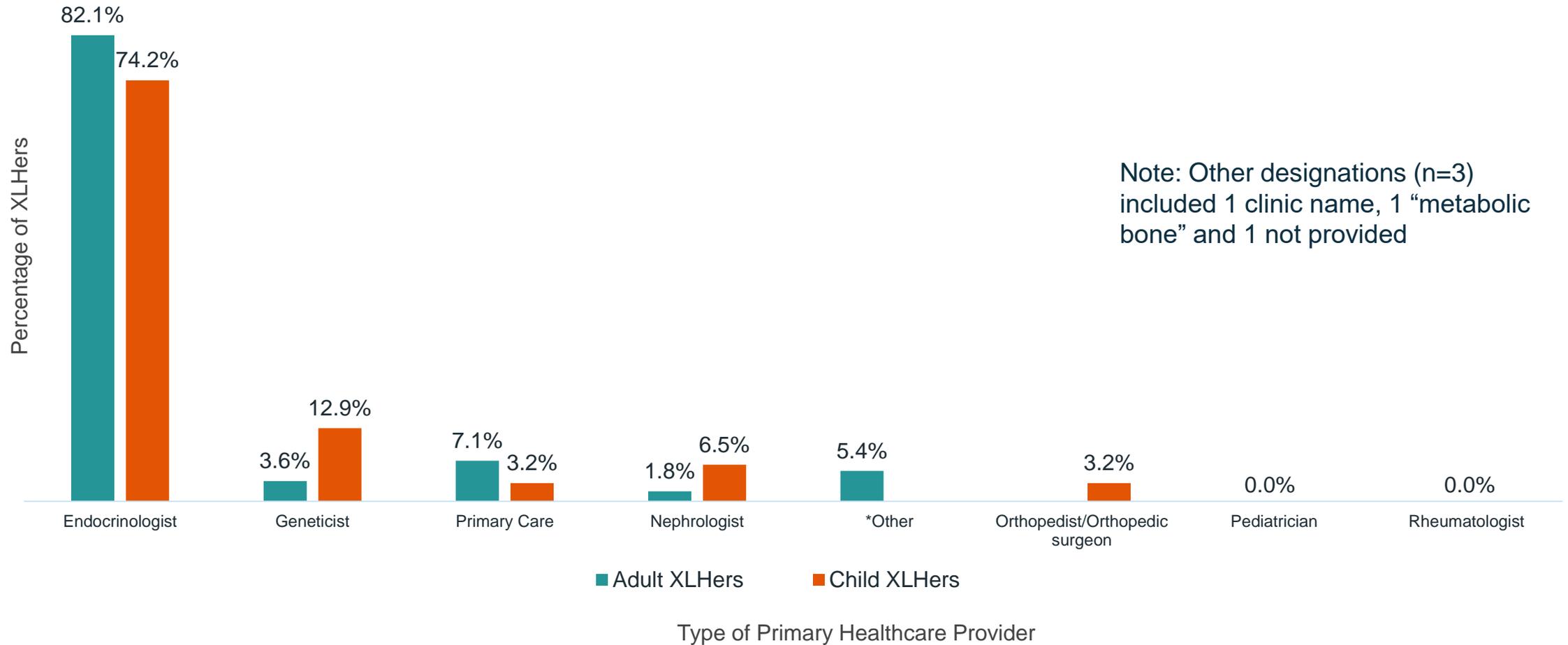
- Adult XLHers rated scheduling process and distance traveled somewhat lower than caregivers representing child XLHers

# Access to Expert Quality Care

- XLH Diagnosis
- Main XLH Care Experiences
- Dental Care Experiences
- Care Transition Experiences

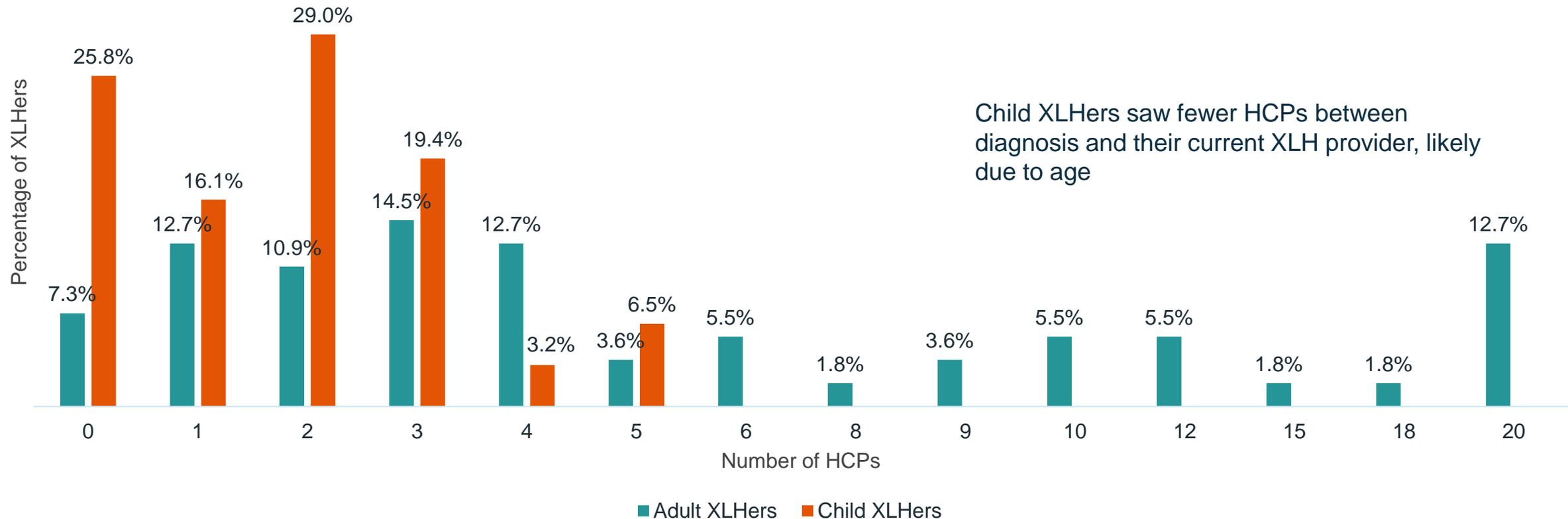
# XLHers are primarily managed by an endocrinologist for their XLH

## Types of HCPs Seen by Respondents for as Primary XLH Provider



# Adult XLHers saw as many as 20 HCPs between diagnosis and their current provider

## Number of HCPs Seen Between Diagnosis and Current XLH HCP



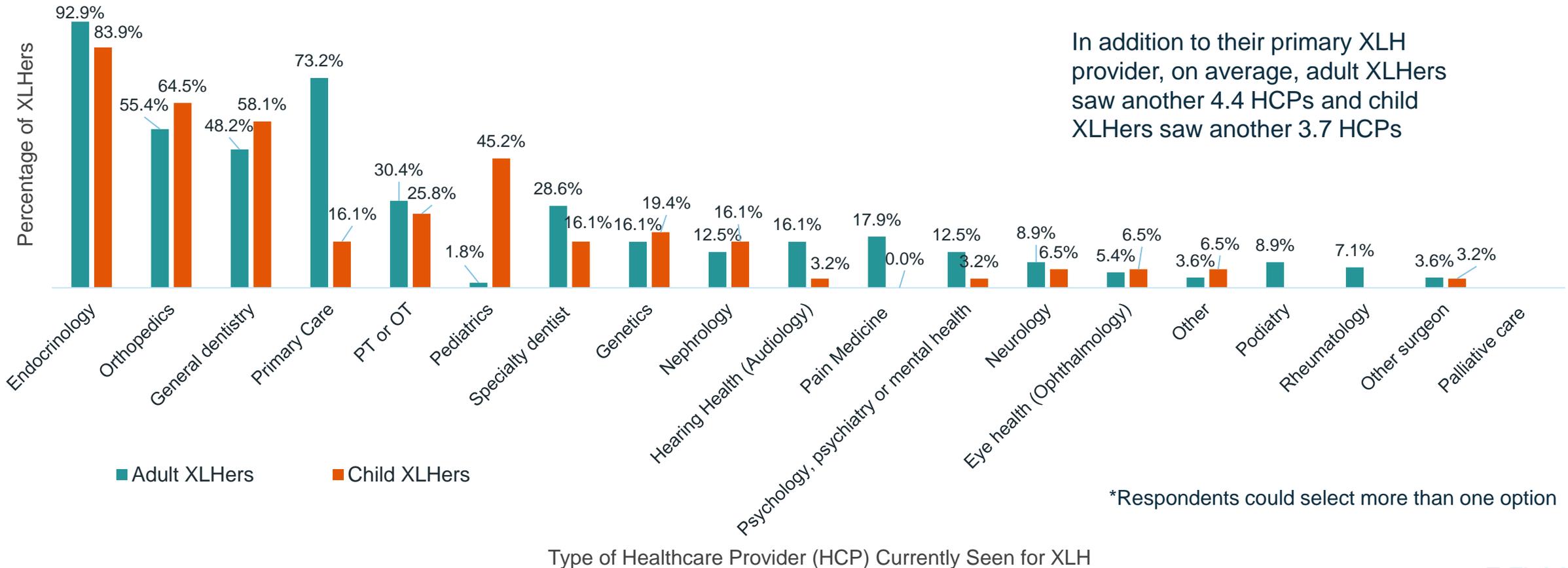
N=87, 56 adult XLHers, 31 child XLHers

QX #12: How many physicians were seen by you/your child between the healthcare provider who diagnosed you/your child and healthcare provider who is the current XLH provider?

# Endocrinology, orthopedics, and general dentistry were the provider types most often involved in XLH care

Adult XLHers frequently saw Primary Care Providers (PCPs), while caregivers of child XLHers saw pediatric providers frequently

## % of XLHers Currently Seeing Specialists\*



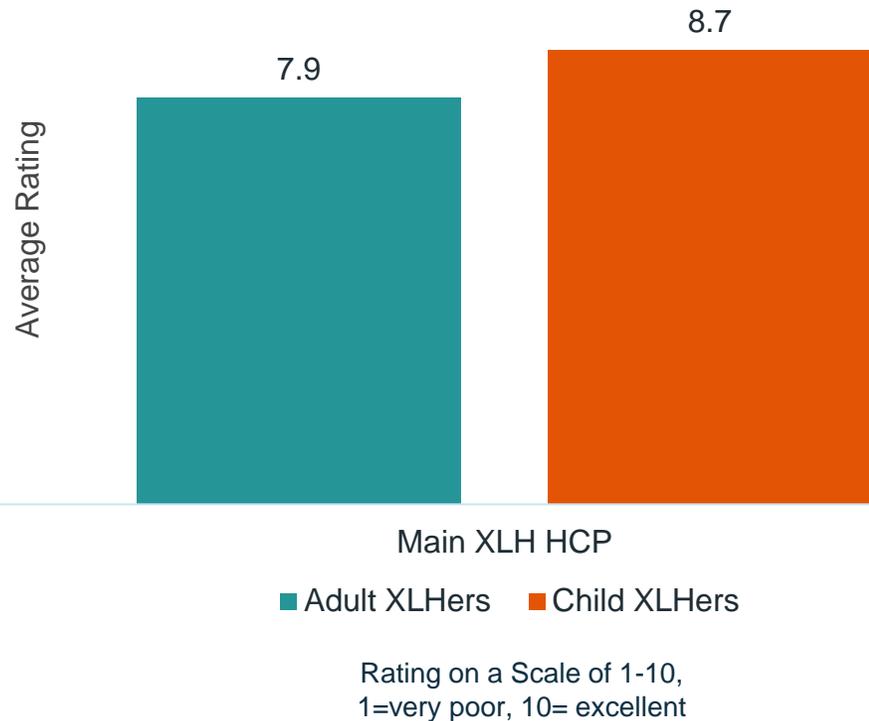
\*Respondents could select more than one option

N=87, 56 adult XLHers, 31 child XLHers

QX #11: Please select all options that describe the specialty of the healthcare providers who you/your child currently see for various issues related to XLH.

# Caregivers of child XLHers rated their main HCP's knowledge of XLH somewhat higher than adult XLHers

### Average Rating of Main HCP XLH Knowledge



#### **Rated main XLH HCP 10/10:**

*My provider now is wonderful. He is very well-versed in XLH and keeps in contact with Yale to keep up-to-date information.*

– Adult XLHer

#### **Rated main XLH HCP 1/10:**

*He just takes care of everyday stuff. My endocrinologist was referred to me... he knows nothing about XLH and doesn't care to learn anything about it. When I bring up an issue, he says my specialty is diabetes. He has another patient with XLH, and he's still not made any effort to educate himself on our disease. There is a doctor in the neuro spine center who led me to this pain management doctor who actually knew the most of any doctor I've ever had but I don't need pain management.*

– Adult XLHer



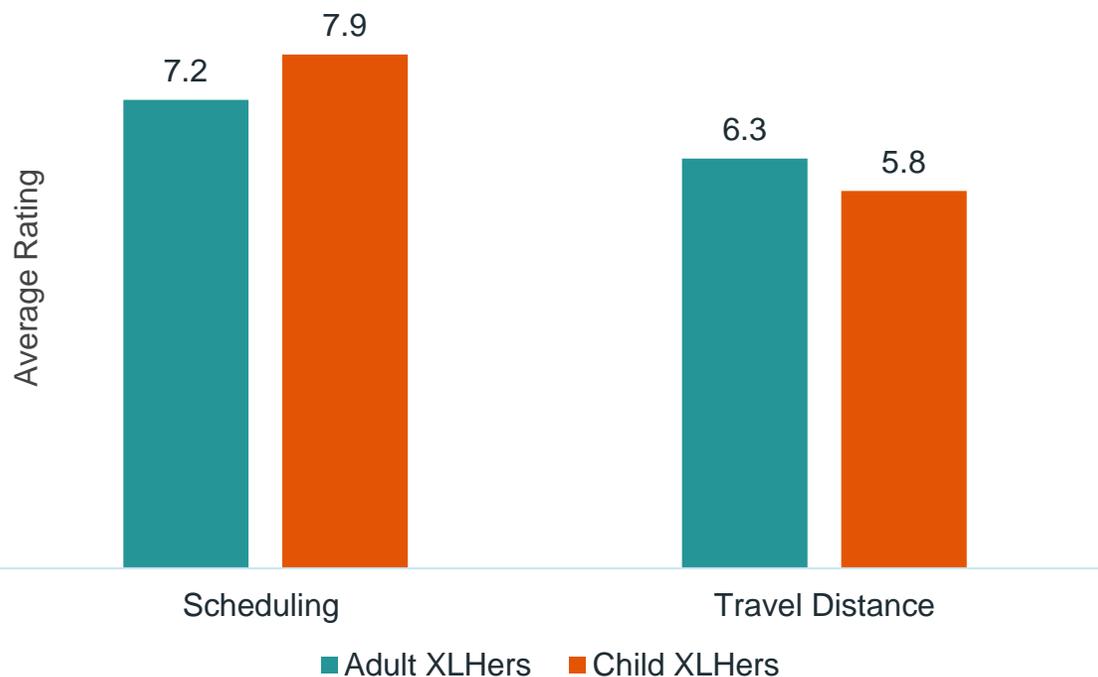
N=87, 56 adult XLHers, 31 child XLHers

QX #13: On a scale of 1-10, with 1 being extremely poor, and 10 being excellent, how would you rate the expertise of each healthcare provider related to their knowledge of XLH?

# XLHers were critical of travel distance to their main XLH provider, averaging 89.5 miles one way with an upper range of 625 miles one way

Both groups rated the scheduling process better than travel distance

**Average XLHer Ratings of Scheduling and Travel Distance to Main XLH Provider (1-10 scale)**



Rating on a Scale of 1-10, 1=very poor, 10= excellent

## Distance and Travel Time to Main XLH Provider

Travel to Main XLH HCP:	Avg Miles One Way (Range)	Avg Hours One Way (Range)
Adult XLHers	89.3 (1-625)	1.6 (0-2.5)
Child XLHers	89.8 (2-500)	1.7 (0-7)

Rated scheduling of main XLH HCP 1/10: "Never answers calls and when in appointments doesn't listen to any concerns"  
 "Can only get an appointment 8 months from when you need it"

Rated scheduling of main XLH HCP 10/10: "Easy to schedule one time a year appointment"  
 "My child's main healthcare provider has great knowledge and communication"

N=87, 56 adult XLHers, 31 child XLHers

QX #14 and 15: On a scale of 1-10, with 1 being extremely poor, and 10 being excellent, how would you rate the process of scheduling a visit with the main healthcare provider for XLH?

## Examples of worst travel or wait-time experiences from survey respondents

“

*Having to wait 6 months from a nephrologist to endocrinologist for XLH related stuff, that was the longest. **My first mental health therapist had an 8 month wait** so I waited that time to see her.*

– Adult XLHer

*I think my worst was when I had to see an endocrinologist in Denver, **had to deal with the horrible traffic to get there, waited 2.5 hours past my time to get in and I got 10 minutes with the doctor** and had to get back in the car- driving much later than I wanted to because of how late my appointment went.*

– Adult XLHer

*Getting there and **having to wait probably a couple hours for the doctor's appointments and the blood work, if they want to do x-rays or ultrasounds it takes a lot longer**, every time we go and have that done we are there all day.*

– Caregiver of child XLHer

*We went to [a] metabolic clinic... and it was so full that we couldn't get in the parking garage, had to get out on the road and walk across. Couldn't get on the elevator - got into the appointment 5 minutes late and I had heard people being told they could not see the doctor after being late - they don't care. Thankfully, we got in.*

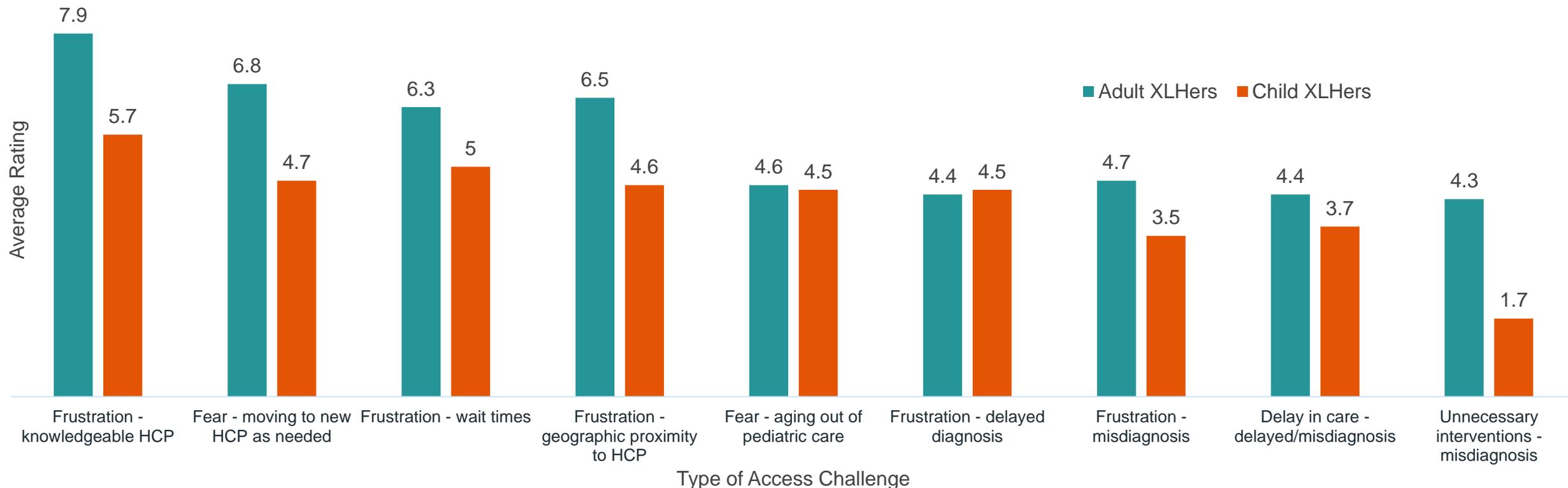
– Caregiver of child XLHer

”

# When asked about challenges in seeking a diagnosis and medical care, the issue rated most highly among XLHers was frustration in finding knowledgeable HCPs

Fear of moving to a new HCP as needed was second for adult XLHers, and frustration regarding wait times was second for caregivers of child XLHers

## Average Ratings of XLH Care Access Challenges (Scale 1-10)



Rating on a Scale of 1-10,  
1=have not experienced, 10= experienced significantly

N=87, 56 adult XLHers, 31 child XLHers

QX #27: On a scale of 1-10, with 1 being "have not experienced" and 10 being "experienced significantly", please rate your experience with each issue related to accessing a diagnosis and medical care related to XLH

## When discussing their frustrations around accessing knowledgeable care, the burden of having to educate providers emerged as a key theme

*“My entire life I was bounced from one doctor to another who didn't know what they were talking about. Even with specialists, they say 'adults don't need [treatment],' which is frustrating.*

*–Adult XLHer*

*I would love to be able to find a different endocrinologist and am willing to travel. **My current doctor is young, has no desire to learn anything,** and I only get to spend about 5 minutes with him”.*

*–Adult XLHer*

*It's just been really challenging ever since I transitioned to adulthood. For the past 5 years, I had a good endocrinologist and he retired. **A lot of times, you are the one educating them.***

*–Adult XLHer*

*Having to find the care on my own is challenging and you have to come **armed with information** for whoever you do see.*

*–Caregiver of Child XLHer*

*Any time I tried to get care it's been difficult, especially with endocrinology. When I lived in a small town, and I had to travel hours for even a little bit of knowledge. **When we moved to a new state, I tried to find one in my city and couldn't get them to take my case.** That was really frustrating, and I had to travel 75 miles each way to the doctor who hadn't ever heard of XLH.*

*–Adult XLHer*

# XLHers who rated their current XLH provider's knowledge of XLH low (1-5 on 10-point scale) cited a general lack of XLH knowledge, lack of a 'holistic approach' to care, and an unwillingness to learn about XLH in interviews

## Top Reasons Cited for Low Ratings of Current XLH Providers



*This provider is pretty new to me, the one I originally saw retired, and he knew a lot more. I am seeing someone who only has 2 XLH patients. I wish she had a **better understanding of “whole system things”** and could more clearly articulate things I can do to help with pain management, and knowledge about how the disease will progress as I age. She is just generally lacking context.*  
–Adult XLHer

*My HCP doesn't know anything about XLH. The infusion center walks her through my renewal each year, she understand and listens, but her knowledge isn't anything. But she doesn't argue and does what I need her to do, so that is helpful.*  
–Adult XLHer

## Participants who experienced minimal frustration in finding knowledgeable healthcare providers were not probed about the subject; unprompted, some mentioned their providers were experts or had other patients with XLH

For the one adult (out of 56) who noted no frustration, there is upcoming stress of their current doctor leaving

“

*My current endocrinologist is amazing, but she is leaving soon, so now I am petrified that her... (successor) won't be as good.*

– Adult XLHer

*The endocrinologist who diagnosed my child happened to be an expert in XLH and we still go there for care.*

– Caregiver of child XLHer

*The nephrologist who diagnosed my child did everything that they could. She was the youngest XLH case they had so there was a bit of uncertainty at first but not much and we continue to receive care there.*

– Caregiver of child XLHer

”

# 32 XLHers rated their current provider a 10/10 for their knowledge of XLH; interviews revealed factors behind high ratings included involvement in XLH research, the HCP's training, and the XLHer's familiarity with the HCP

4 caregivers noted the diagnosing physician was so knowledgeable they decided to continue care with them

## Top Reasons Cited for 10/10 Ratings of Current XLH Providers



*“She treats a lot of other kids with XLH, so she is **very familiar**. She also **studied with one of the premier XLH doctors** and is very knowledgeable.”*  
–Caregiver of child XLHer

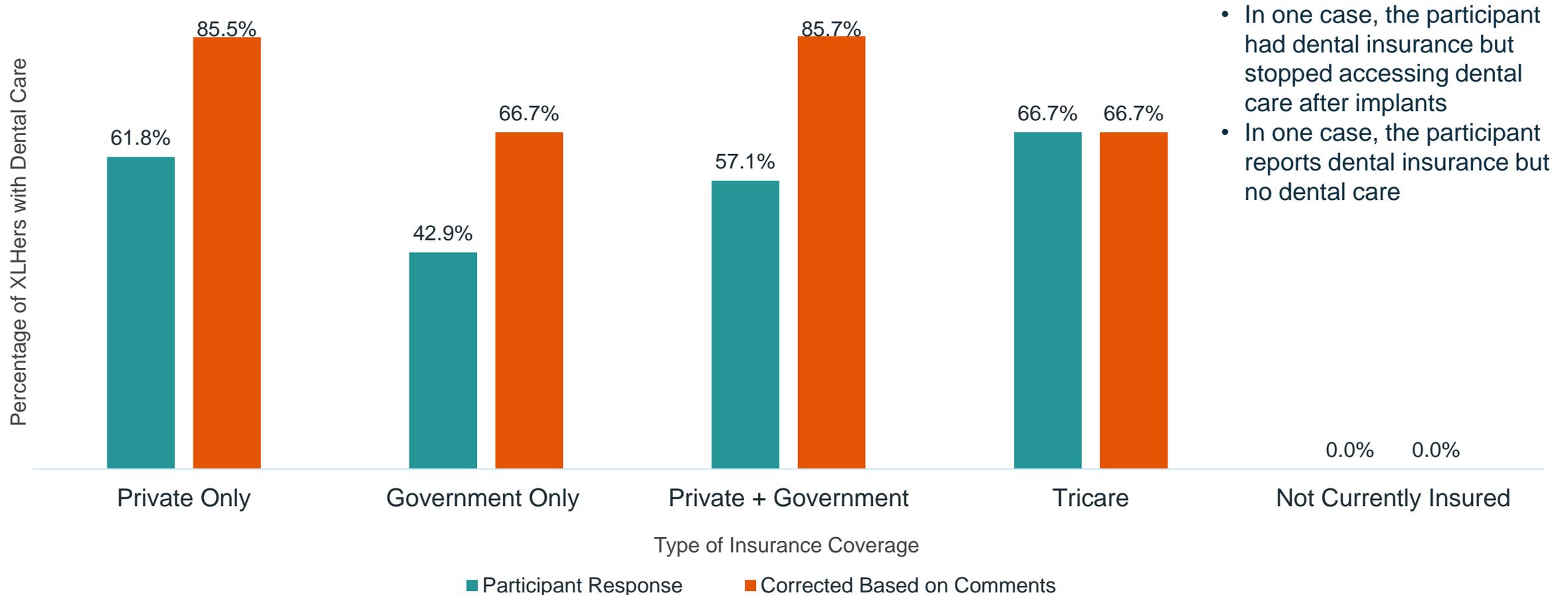
*He specializes in XLH and is very **knowledgeable about the disease** and **has other patients**. He makes sure that I get my renal ultrasounds and is **ahead of things** sometimes. If something is hurting, he helps me get things to manage it.”*  
–Adult XLHer

# Access to Expert Quality Care

- XLH Diagnosis
- Main XLH Care Experiences
- Dental Care Experiences
- Care Transition Experiences

# Information provided by respondents about dental care was inconsistent; certain respondents who reported that they did not have dental care reported out of pocket expenses. When data were *corrected*, it appears that 79.3% (not 56.3%) actually had dental care

## Corrected Percent of Patients with Dental Care, By Insurance Type



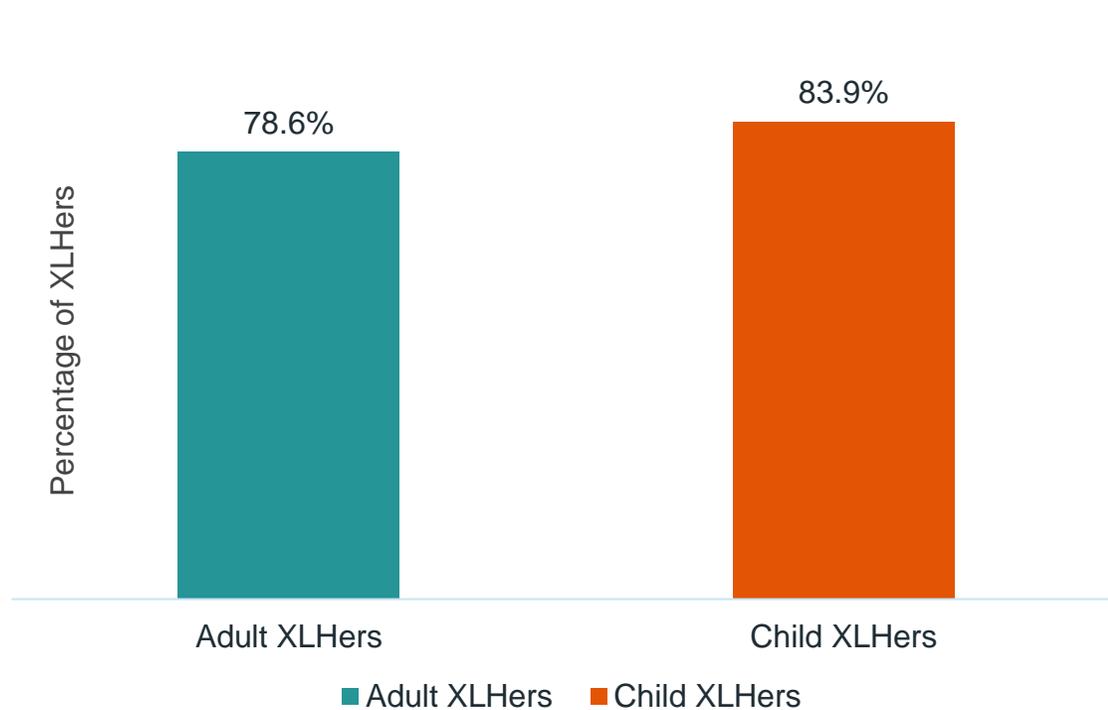
N=87, 56 adult XLHers, 31 child XLHers

QX #11: Please select all options that describe the specialty of the healthcare providers who you/your child currently see for various issues related to XLH

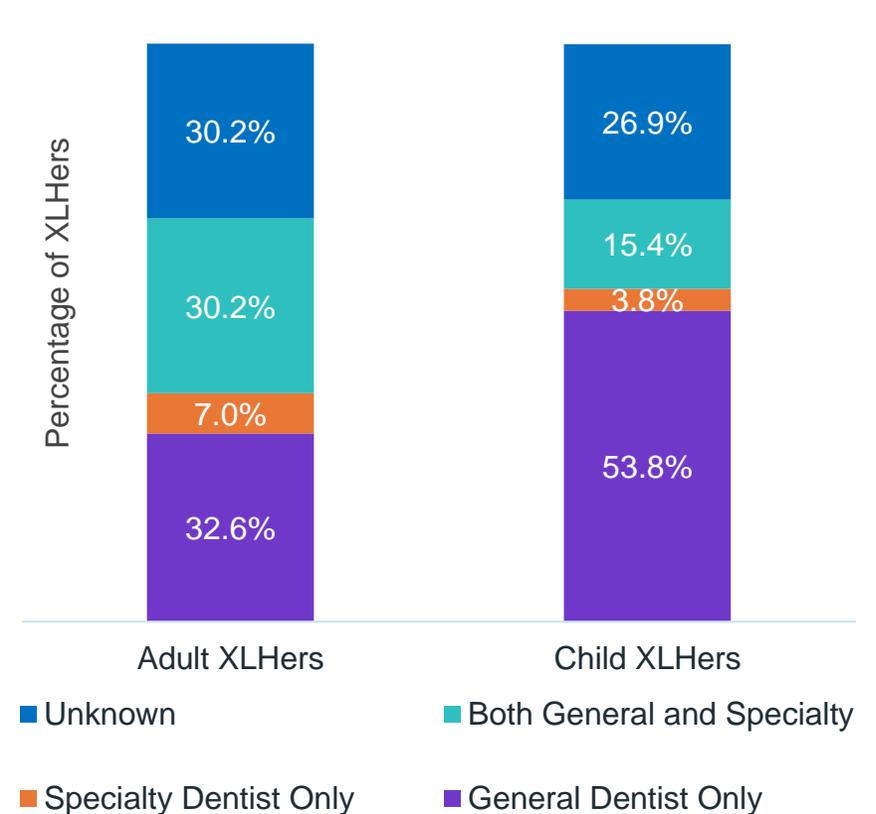
\*General dentist and specialty dentist were both options within this list

**When data were *corrected*, it appears that 78.6% of adult XLHers and 83.9% of child XLHers had a dentist, most often a general dentist (though survey data and inconsistent reporting left some responses unknown)**

**Percent of XLHers with Dental Care**



**Type of Dentist Seen**



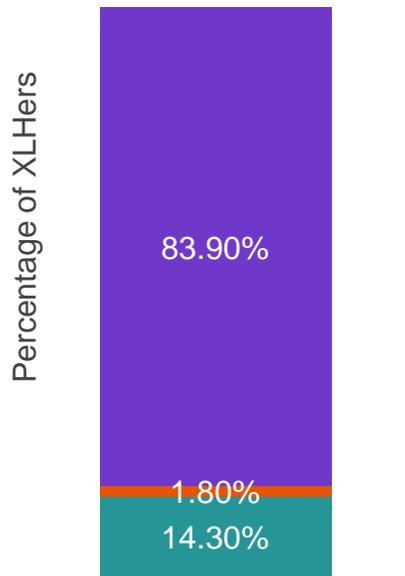
N=87, 56 adult XLHers, 31 child XLHers

QX#11: Please select all options that describe the specialty of the healthcare providers who you/your child currently see for various issues related to XLH

\*General dentist and specialty dentist were both options within this list

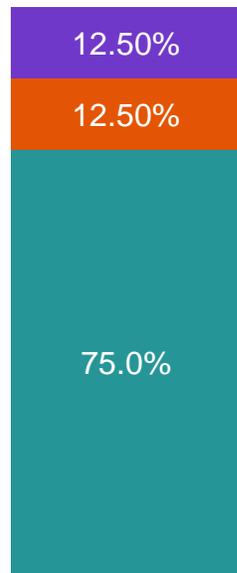
# Majority of XLHers with a current, consistent dentist say their dentist is receptive to learning about XLH

How Dentist was Found



- Found on Own
- Referred
- Not with Current Dentist

Dentist's Receptiveness to Learning about XLH



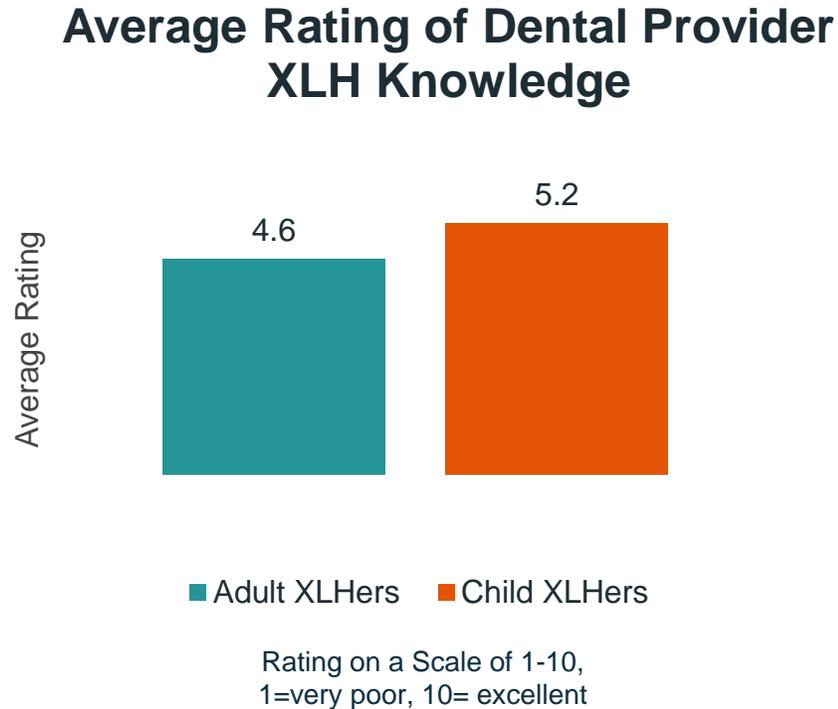
- Unclear if Receptive
- Not Receptive
- Receptive

“95% of the time dentists **don't know what XLH is**, I wouldn't say I've met a dentist who is excited to learn about it. My child hasn't had dental issues, so the dentists haven't seen a need to learn about it. The dentist we just went to specializes in implants, so she will be ready for that issue” (Unclear if receptive to learning about XLH).  
 --Caregiver of Child XLHer

The dentist **didn't know about XLH when we first went**, but the more that we go over the years, she is more understanding of what is going on with my child's teeth, the decay issues and soft teeth, and she relates it back to XLH” (Dentist is receptive to learning about XLH).  
 --Caregiver of Child XLHer

Dentists often are listening but **aren't really hearing you**. You become a 'professional patient' and learn how to educate them. I don't have anyone consistent- I am currently in the market for a dentist and have a good referral, so I am **about to go to a new one**. I haven't been to them yet.  
 --Adult XLHer; person is currently without a dentist

# On average, XLHers rated their dentists' knowledge of XLH quite low, on a scale of 1-10



**Rated dentist 10/10:**

*I used to go to one dentist, and they said they weren't comfortable treating me. I found a new one and told them I had XLH and wasn't a typical XLH case. They had read every article and knew a lot about it when I went there.*

--Adult XLHer

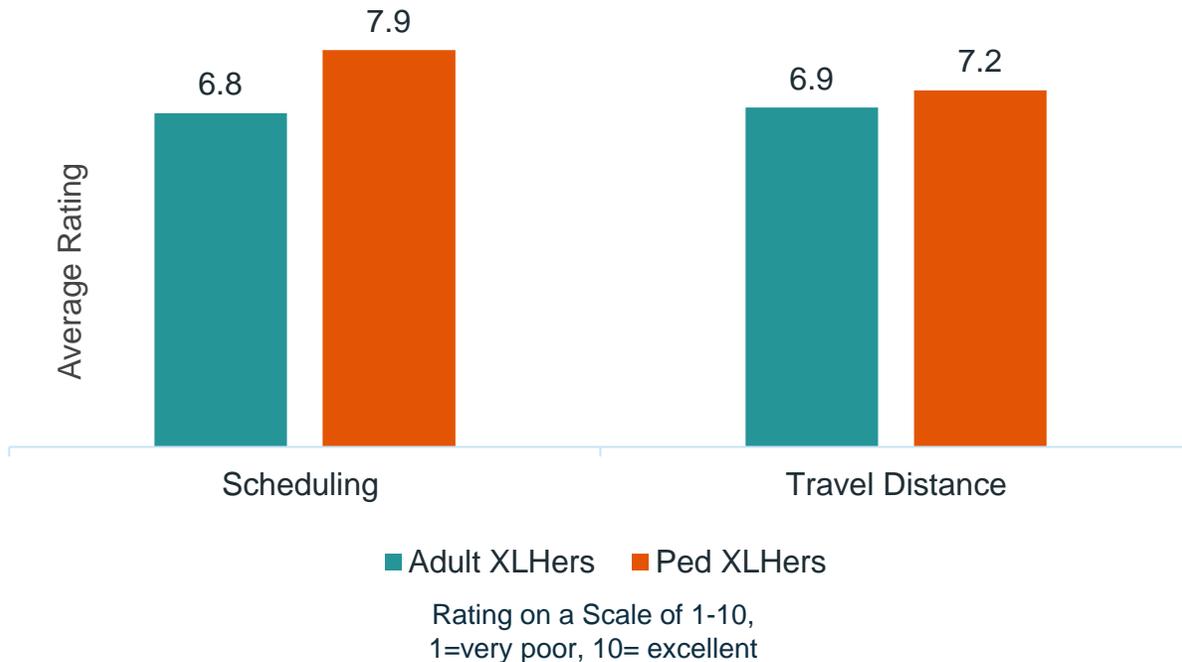
**Rated dentist 1/10:**

*My dentist has no idea about XLH, no clue. They are willing to learn for the most part, but they don't seem interested in caring.*

--Adult XLHer

# On average, XLHers travel 12.6 miles one way to their dental care provider

Average XLHer Ratings of Scheduling and Travel Distance to Dental Care Provider (1-10 scale)



Distance and Travel Time to Dental Care Provider

Travel to Dental Care Provider	Avg Miles One Way (Range)	Avg Hours One Way (Range)
Adult XLHers	14.1 (0-150)	0.8 (0-2.3)
Child XLHers	9.5 (0.5-30)	0.2 (0-0.75)

**“** *Rated scheduling of dental care 10/10:*

*Easy to schedule and easy to get in.*

*I never have an issue scheduling an appointment for my child.* **”**

n=70, 44 adult XLHers, 26 child XLHers

QX #14 and 15: n a scale of 1-10, with 1 being extremely poor, and 10 being excellent, how would you rate process of scheduling and the travel distance to see the dental care provider?

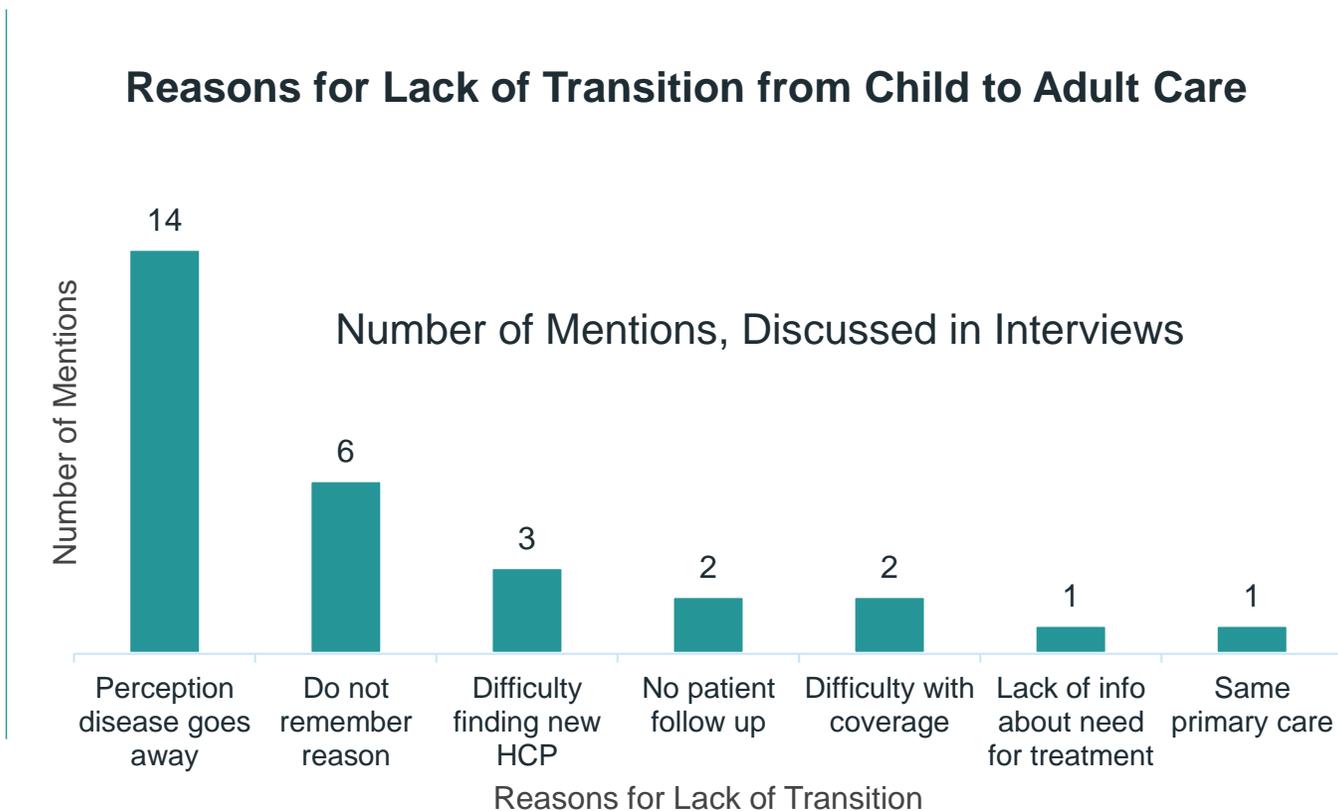
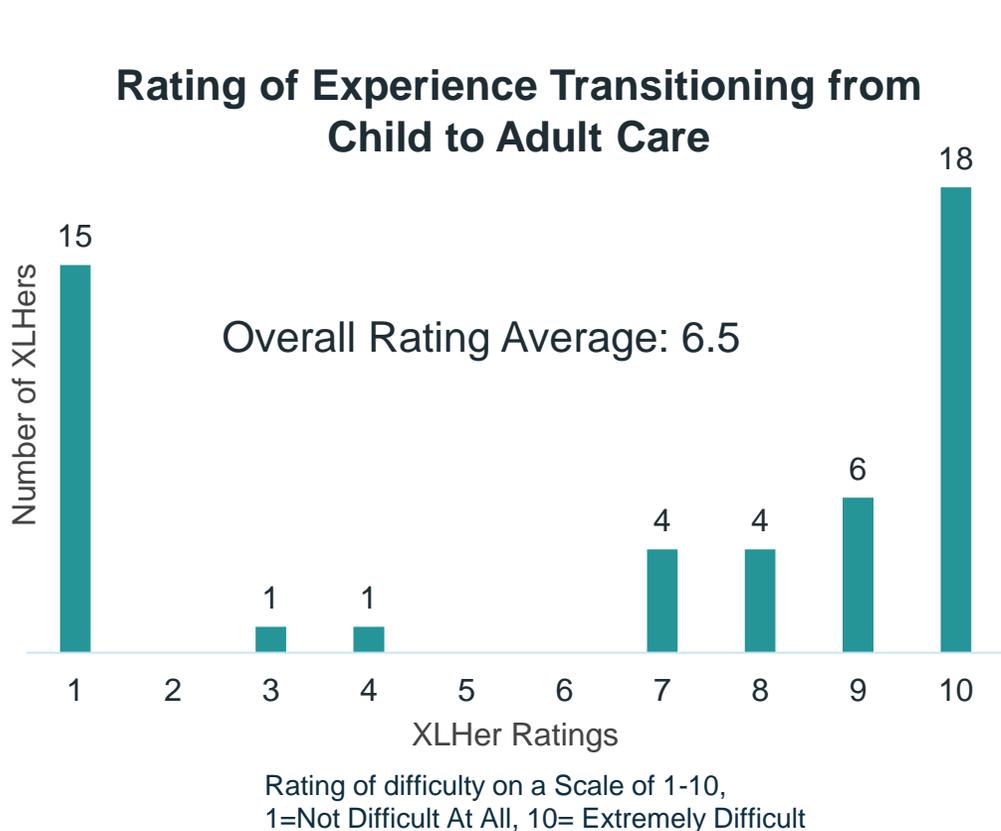
# Access to Expert Quality Care

- XLH Diagnosis
- Main XLH Care Experiences
- Dental Care Experiences
- Care Transition Experiences

# Being told “the disease would go away” remains the top reason for not experiencing a transition from pediatric to adult care among XLHers diagnosed before age 18

Among the 49 adult XLHers diagnosed before age 18, 47 (95%) did not undergo a true transition of care

Note: This slide excludes data from 6 adult XLHers diagnosed after age 18



n=49 adult XLHers

QX 30 and 63: At the time you were transitioning from pediatric to adult care, how difficult was it to find a new doctor on a scale of 1-10 with 1 being not difficult at all and 10 being extremely difficult?

If participant is age 18 or older, if <18, skip: Tell me more about transitioning from pediatric to adult care...what could have gone better or been done to improve the transition?

## The 2 adult XLHers reporting a smooth transition to adult care were either able to stay in the same healthcare system or received good communication from their care team

“

*From what I remember I went right through and I didn't have too much trouble. My endocrinologist was within the same system that I was already in. I only had to go up one floor and also had no copays, because I don't pay for doctor visits or lab work as long as it stays in that system.*

*– Adult XLHer, current age 57, transitioned in 1984*

*They were very good at communicating once I switched over from pediatric to adult care.*

*– Adult XLHer, current age 21, transitioned in 2021*

”

## Many adult XLHers who did not undergo a transition of care were told that they had ‘grown out’ of the disease, and often felt they had nowhere to turn for care

*I wish they would've had packets of information for those aging out and transitioning to adult care. **I thought I was fixed** and didn't really learn enough to know. Having the emphasis on how important it is would have been a big help to not let things keep going downhill.*

*At 18, I stopped seeing my childhood specialist and they didn't know where to send me; there was nobody else and **I was on my own**. At 18 I didn't take it seriously, so I never went out and found a specialist. From 18-31, I wasn't being treated, I didn't know anybody with the disease and had no direction. When my daughter got diagnosed, I started googling and getting care again – I was about 32. It would have helped if they knew who to send me to or somebody in charge of advocating for children, that's what I do now - making sure no kid goes without care for transitioning from childhood to adult. I wish that was there for me because just being informed and having the information to transfer you is huge.*

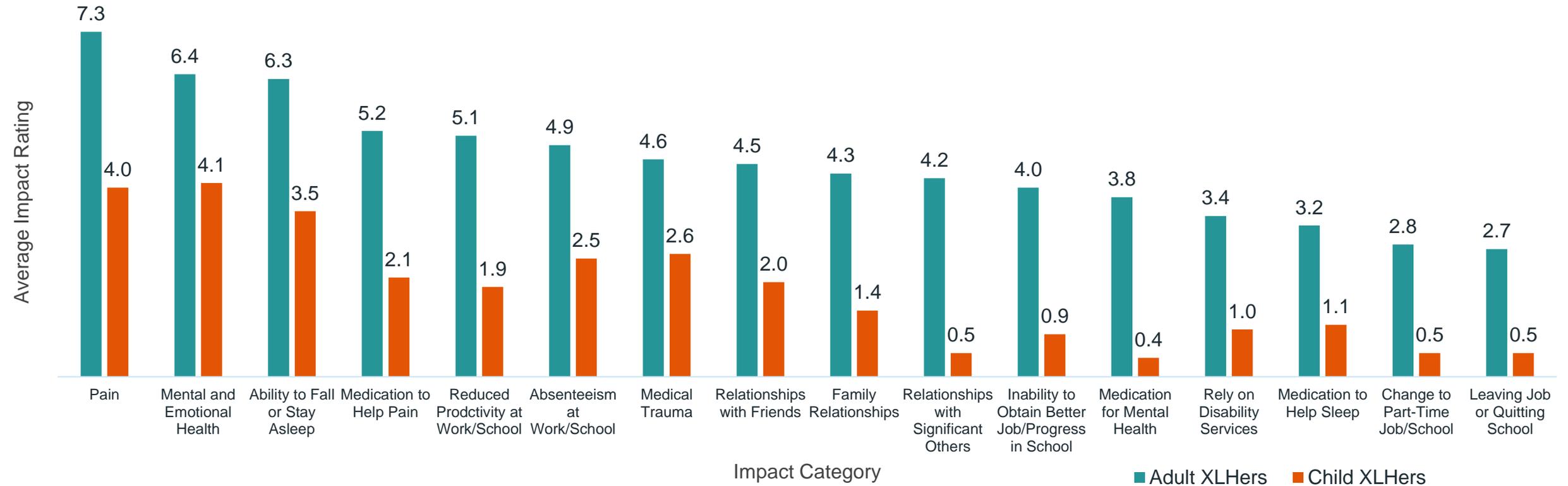
*I aged out at Shriner's when I became an adult, and I didn't have any other doctors I could go see. **I didn't have anywhere to turn... and they didn't help me find a new person**. There was a long gap, and the next 10 years was sporadic care here and there until maybe 2016 when I found information online.*

# Physical and Psychosocial Impact

- Physical Impacts
- Psychosocial Impacts

# When psychosocial impacts were analyzed across all participants, pain was rated as the highest impact followed by mental/emotional health and the ability to stay asleep

Impact of XLH on Adults and Children Living with XLH in the Last Year (1-10 scale)



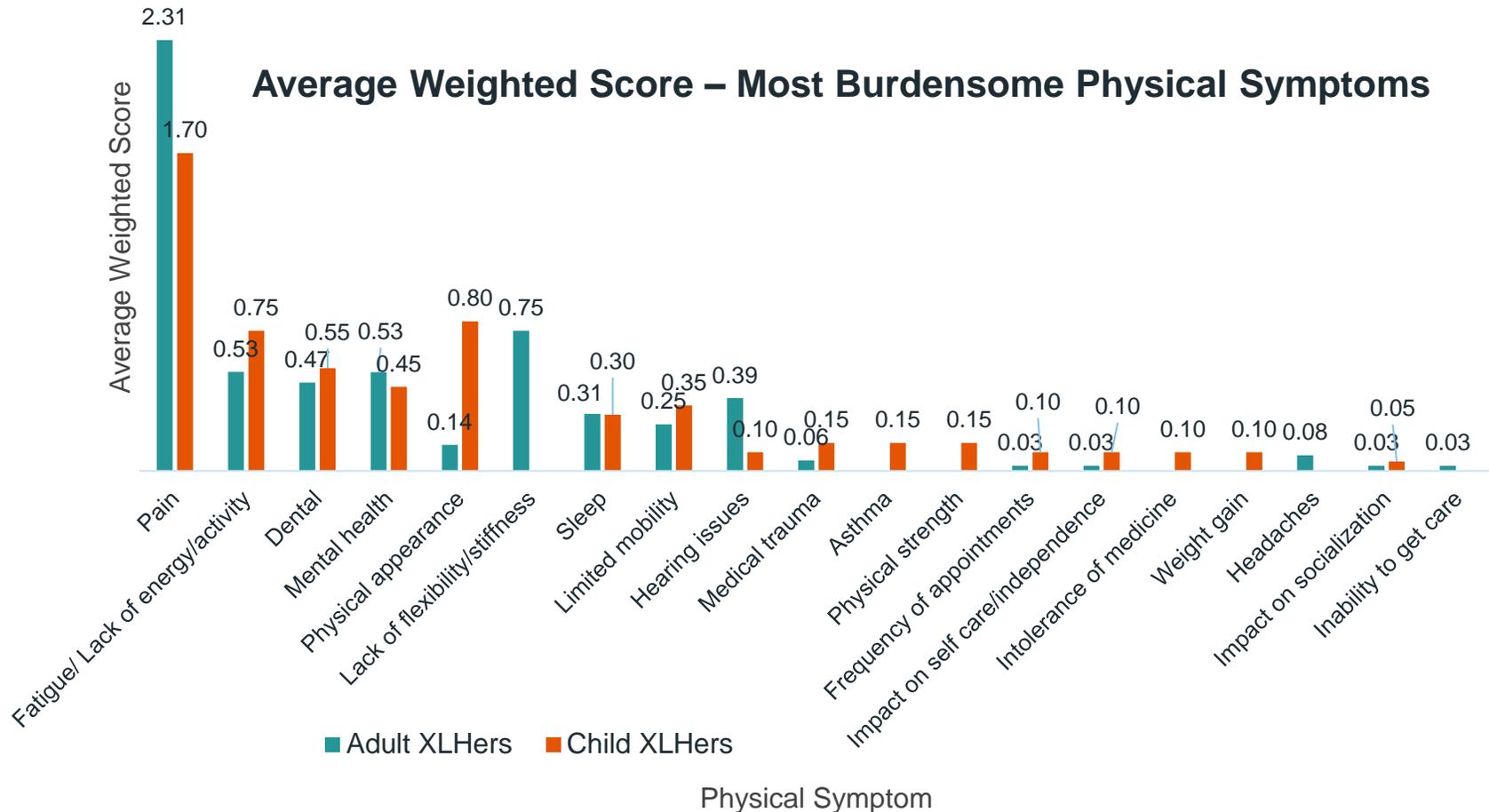
Average Rating on a Scale of 1-10, 1=no impact, 10= extreme impact

N=87, 56 adult XLHers and 31 child XLHers

QX #16: On a scale of 1-10, with 1 being "no impact / did not experience" and 10 being "extreme impact / experienced significantly", please rate the impact that XLH has had on you/the person diagnosed with XLH in the last year in each of the following areas

# Both groups identified pain as the most burdensome physical symptom

Adult XLHers reported lack of flexibility/stiffness as the second most burdensome, while caregivers of child XLHers reported physical appearance



## Methodology for Analysis:

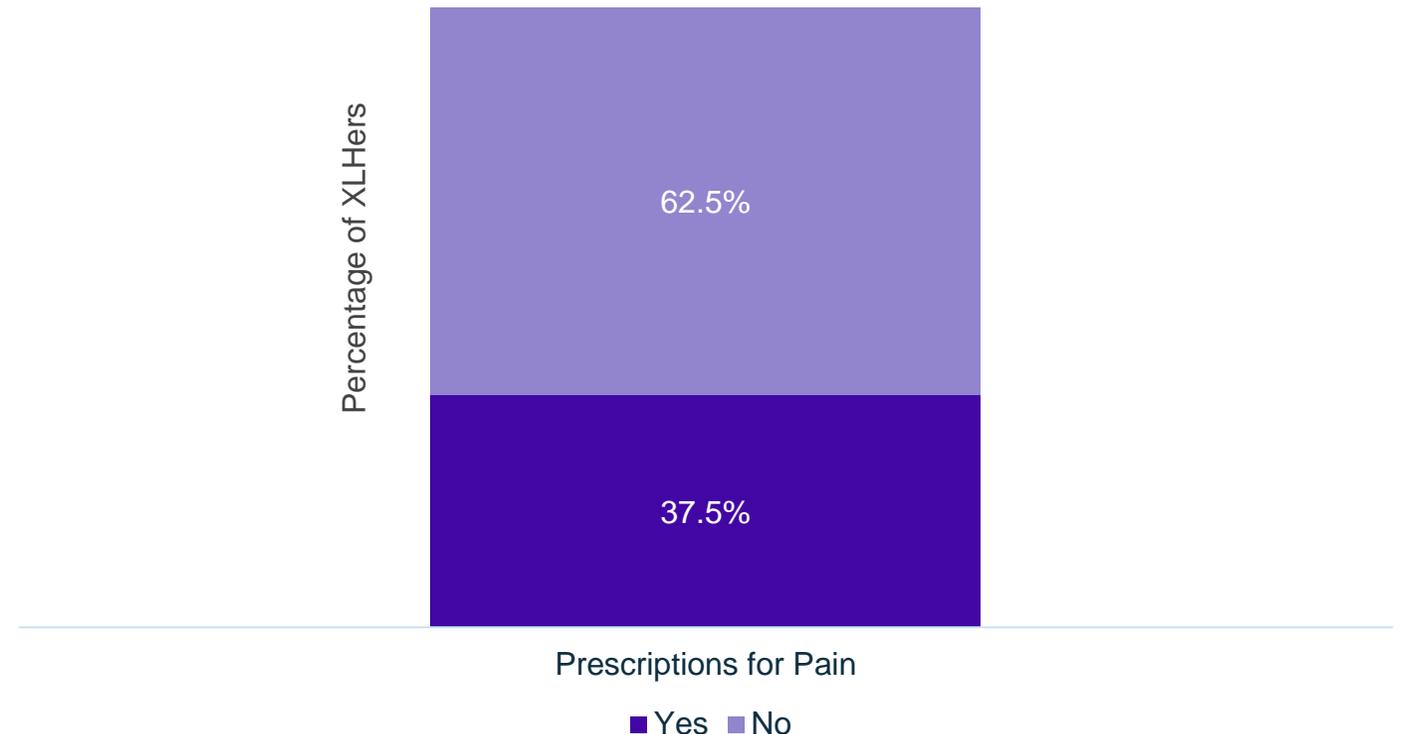
- Participants were asked in the interview to provide their top 3 burdens
- The #1 most important was assigned three points, the second assigned two, and the third assigned one point
- This means each respondent had a possible 6 points to assign – please note that some respondents did not have three issues to list (e.g. caregivers with small children)
- Total points were divided by the number of respondents in that category (Adults = 36, Children = 20): this results in an average weighted score for each item

# Just under 1/3 of adult XLHers were prescribed medications for pain, but no child XLHers had prescriptions for pain

## Pain Medications

- Meloxicam (3 mentions)
- Other treatment (3 mentions)
- Ibuprofen 800 mg (2 mentions)
- Oxycodone (2 mentions)
- Oxycodone acetaminophen (2 mentions)
- Tramadol (2 mentions)
- Celecoxib (1 mention)
- Cyclobenzaprine (1 mention)
- Carisoprodol (1 mention)
- Gabapentin (1 mention)
- Methadone (1 mention)
- Morphine ER and IR (1 mention)
- Naproxen sodium (1 mention)
- Hydrocodone (1 mention)
- Pain pump (1 mention)

## Adult XLHers Prescribed Pain Medication



N=87, 56 adult XLHers and 31 child XLHers

QX #22: Are you/your child currently prescribed FDA-approved medications (for example, medications not available over the counter or medications that require a prescription from your healthcare provider) to help manage pain or depression?

## Illustrative quotes regarding the impact of pain

“

*For as long as I can remember, I have dealt with pain. It stands out more when I don't have pain rather than when I do, it's just a chronic thing that affects different areas. You get used to it, but it's always there. I am cautious with my pain too. It always concerns me, I am wondering what the pain is from, if is it bone pain or a calcium deposit... wondering what it is and when to go to the doctor. It can be debilitating.*

– Adult XLHer

*Pain impacts my stamina. There is a lot of bone and joint aches and pains, it comes and goes. I do have things I do (like exercises) that make me feel better, stretching, a red light used for aches and pains, and hot baths. I also have a prescription that I try not to take. I do turmeric and CBD oil, and other ointments to try to manage it. I can go weeks without any issues and then **suddenly I stand wrong or something and I'll hurt again.***

– Adult XLHer

*After playing hard all day, it will come up - **her legs will hurt.** She's very active so it does come up. There were maybe a couple times a month before her medication was adjusted and now it is less often than that.*

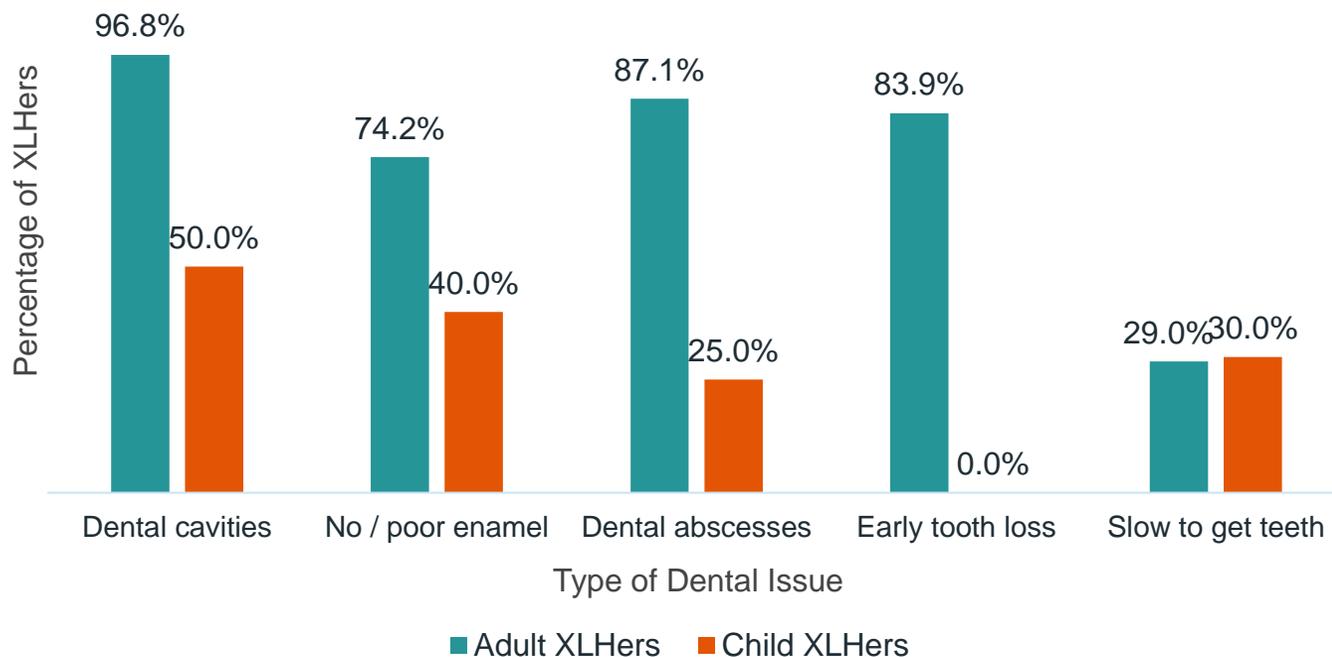
– Caregiver of child XLHer

”

# Adult XLHers more frequently reported dental issues; cavities were the most common issue in both age groups

Issues noted by adult XLHers (abscesses and early tooth loss) were more relevant to having adult teeth

### Dental Issues Reported by XLHers and Descriptions



Number of adults with at least 1 dental issue: 97.2%  
 Number of children with at least 1 dental issue: 75.0%  
 Average age of presentation for dental issues for adults: 11.4 years  
 Average age of presentation for dental issues for children: 4.3 years

#### Dental cavities

- 9 mentioned “a lot of cavities”
- 8 mentioned “a few cavities”
- 8 noted cavities “in childhood”

#### Enamel

- 24 mentioned ‘poor enamel’
- 2 mentioned ‘no enamel’

#### Abscesses

- 8 mentioned “a lot of abscesses”
- 3 noted “spontaneous abscesses”

#### Early tooth loss

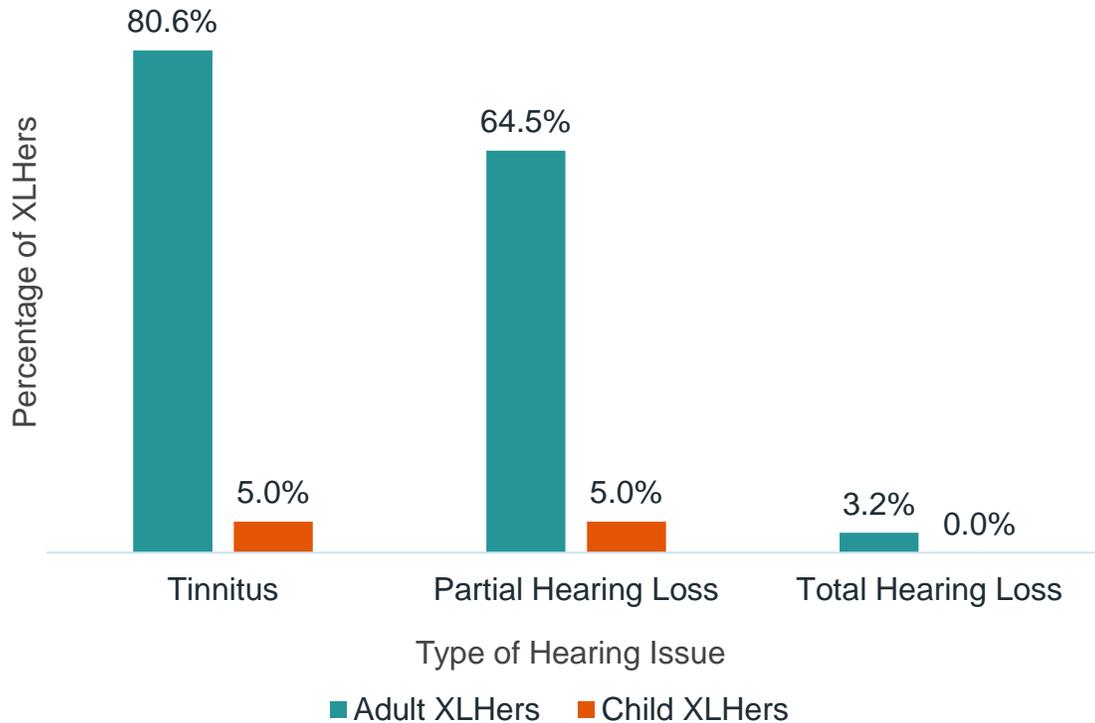
- 8 mentioned losing “majority of teeth”
- 6 mentioned having lost 1 tooth
- 2 noted having dentures

#### Slow to get teeth

- 5 mentioned adult teeth being slow
- 4 noted baby teeth being slow

# As reported by adult XLHers and caregivers of child XLHers, adult XLHers experience hearing issues more frequently than child XLHers, with tinnitus and partial hearing loss as the most common issues

Hearing Issues Reported by XLHers and Descriptions



Number of adults with at least 1 hearing issue: 72.2%  
 Number of children with at least 1 hearing issue: 10.0%  
 Average age of presentation for hearing issues for adults: 34.6 years  
 Average age of presentation for hearing issues for child XLHers: 3.0 years

## Tinnitus

- 11 mentioned “it comes and goes”
- 9 mentioned “it is constant”

## Hearing loss

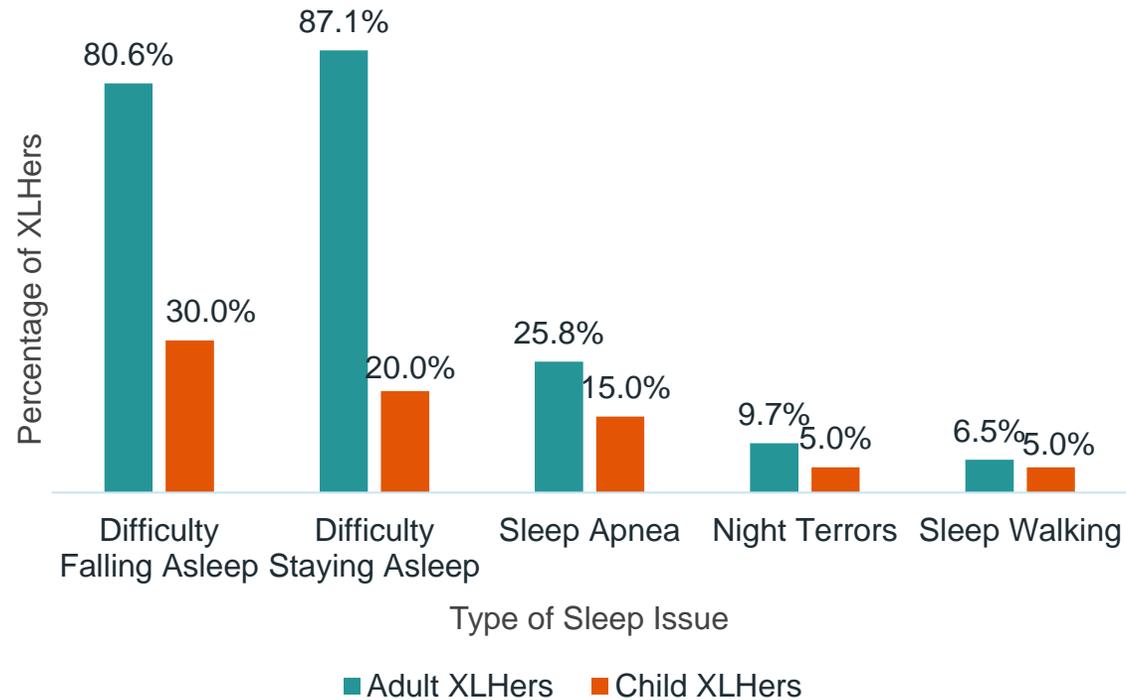
- 8 mentioned hearing loss in both ears
- 5 mentioned hearing loss on one side
- 7 are currently being evaluated for potential hearing loss
- 1 mentioned total hearing loss

## Hearing loss interventions

- 3 wear hearing aids (no cochlear implants)
- 5 mentioned needing to pursue hearing aids (3 due to financial reasons, 2 just haven’t pursued)

# As reported by adult XLHers and caregivers of child XLHers, adult XLHers experience some sleep issues more frequently than child XLHers, with difficulty falling and staying asleep being quite frequent

## Sleep Issues Reported by XLHers and Descriptions



Number of adults with at least 1 sleep issue: 91.7%  
 Number of children with at least 1 sleep issue: 60.0%  
 Average age of presentation for sleep issues for adults: 35.9 years  
 Average age of presentation for sleep issues for children: 4.3 years

### Difficulty falling asleep

- 10 mentions of pain causing issues
- 6 mentions of difficulty on some nights
- 4 mentions of rarely having issues

### Difficulty staying asleep

- 11 mentions of issues related to pain
- 9 mentions of difficulty “most nights”
- 2 mentions each of related to restless legs and bathroom breaks

### Sleep apnea

- 2 mentions of getting a tonsillectomy soon
- 1 mention each of using a c-pap, as a result of weight gain, and “comes and goes”

### Night terrors

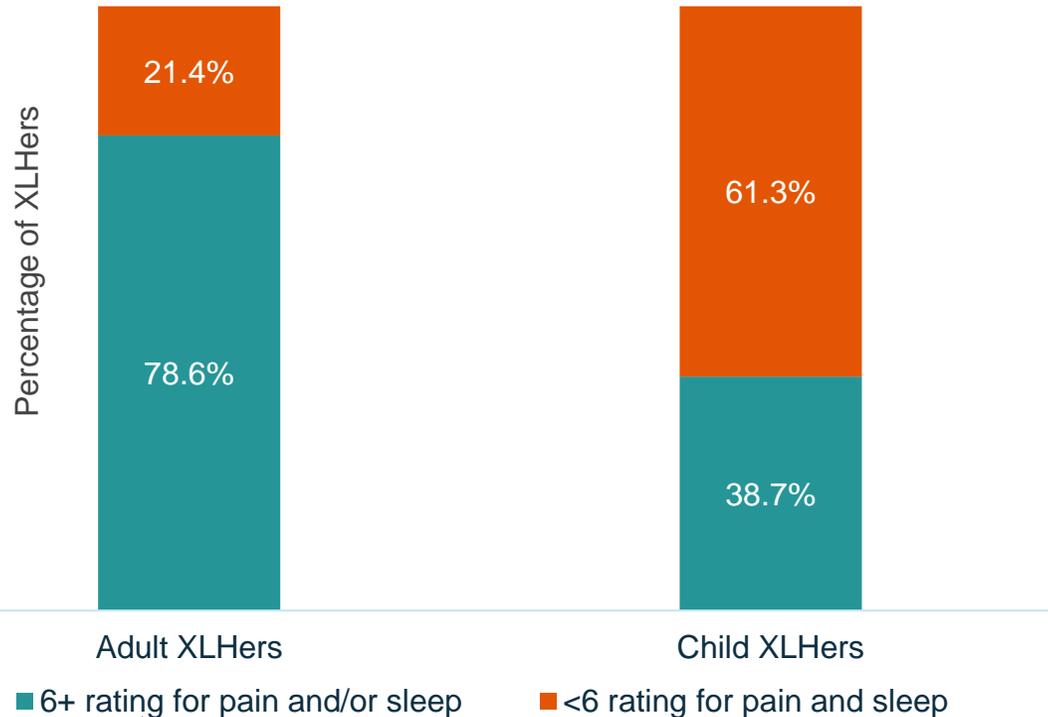
- 3 mentions of night terrors
- 1 intense night terrors once a week
- 1 traumatic night terrors occasionally
- 1 vivid nightmares that are “well-managed”

### Sleep walking

- 1 mention of issues during childhood
- 1 mention of sleep walking happening a “couple times”

# The majority (78.6%) of adult XLHers rated pain and/or sleep issues as a 6 or higher on a scale of 1-10, suggesting that pain and sleep issues are some of the most significant issues affecting physical health, daily activities, and psychosocial issues

## Percentage of XLHers Rating Pain and Sleep Issues $\geq 6$



## Methodology for Analysis

- Though pain and sleep were measured and also discussed in the psychosocial section of the interview guide, it appears that the physical and psychosocial impacts of pain and sleep overlap and also affect other variables.
- To explore this more, an analysis was performed first to determine how prevalent sleep and pain issues were in the study population.

N=87, 56 adult XLHers and 31 child XLHers

QX #16: On a scale of 1-10, with 1 being "no impact / did not experience" and 10 being "extreme impact / experienced significantly", please rate the impact that XLH has had on you/the person diagnosed with XLH in the last year in each of the following areas

# Data suggest a relationship between having pain and/or sleep issues and other impacts, including mental health, work/school, medical trauma, relationships, and relying on disability

Among XLHers who gave a 6+ rating for pain and/or sleep, the average ratings for *other* psychosocial issues were higher in almost every category:



Excludes N/A; Base (N) Size Varies by Item:

Adult XLHers with High Pain/Sleep Rating (6+)	44	42	38	41	42	41	34	41	26	29	23	22
Adult XLHers with Low Pain/Sleep Rating (<6)	12	11	10	11	12	12	11	12	7	9	9	9
Child XLHers with High Pain/Sleep Rating (6+)	10	9	7	8	10	11	6	3	7	4	3	4
Child XLHers with Low Pain/Sleep Rating (<6)	18	16	11	15	17	17	15	8	6	4	9	10

# Physical and Psychosocial Impact

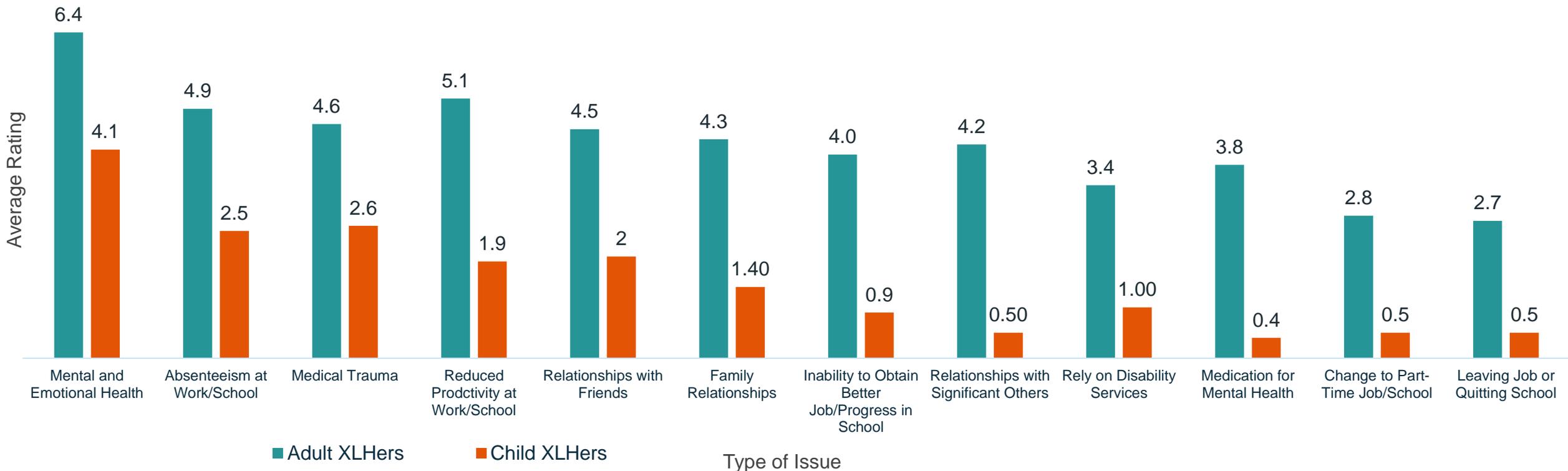
- Physical Impacts
- Psychosocial Impacts

# When pain and sleep issues are removed, mental and emotional health are the most impactful issues in each age group

Reduced productivity and absenteeism at work/school, medical trauma, and relationships with friends were also reported among the top 5 most impactful issues for all respondents

## Average Impact of XLH on Adults and Children Living with XLH in the Last Year

Not including pain and sleep issues



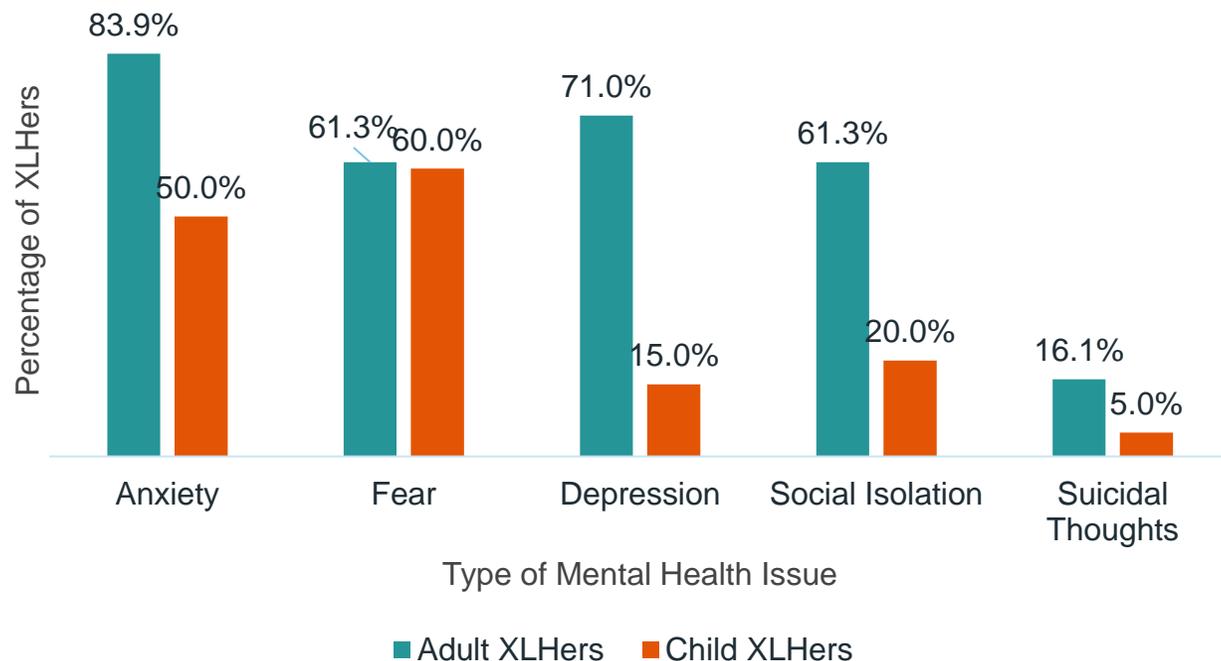
Rating on a Scale of 1-10,  
1=no impact, 10= extreme impact

N=87, 56 adult XLHers and 31 child XLHers

QX #16: On a scale of 1-10, with 1 being "no impact / did not experience" and 10 being "extreme impact / experienced significantly", please rate the impact that XLH has had on you/the person diagnosed with XLH in the last year in each of the following areas

# Mental health issues were prevalent in both age groups, with anxiety and fear being frequently mentioned; adult XLHers also noted a high prevalence of depression and social isolation

Mental Health Issues Reported by XLHers and Descriptions



Number of adults with at least 1 mental health issue: 83.3%  
 Number of children with at least 1 mental health issue: 90.0%  
 Average age of presentation for mental health issues for adults: 30.7 years  
 Average age of presentation for mental health issues for children: 4.6 years

## Anxiety

- 13 mentions of healthcare settings/procedures
- 5 mentions of being somewhere new in general
- 4 mentions of social anxiety
- 3 mentions each of worry regarding progression, stress, and anxiety about falling as contributing factors

## Fear

- 13 mentions of healthcare settings/procedures
- 9 mentions of fear about falling
- 9 mentions of fear about the future / progression of XLH
- 2 mentions each;
  - Socializing
  - Maintaining medical coverage

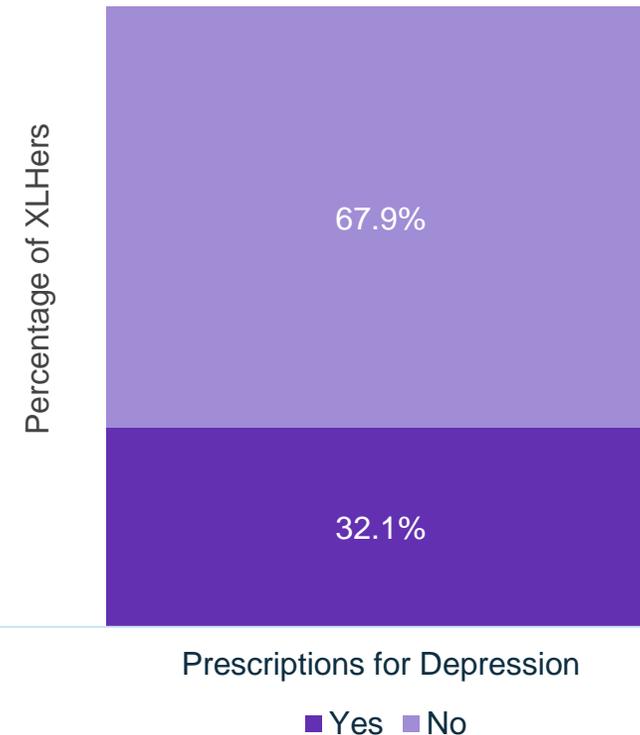
Note: for caregivers of child XLHers, anxiety and fear often overlapped in regard to healthcare settings/procedures

# Just under 1/3 of adult XLHers were prescribed medications for depression, but no child XLHers had prescriptions for depression

## Depression Medications

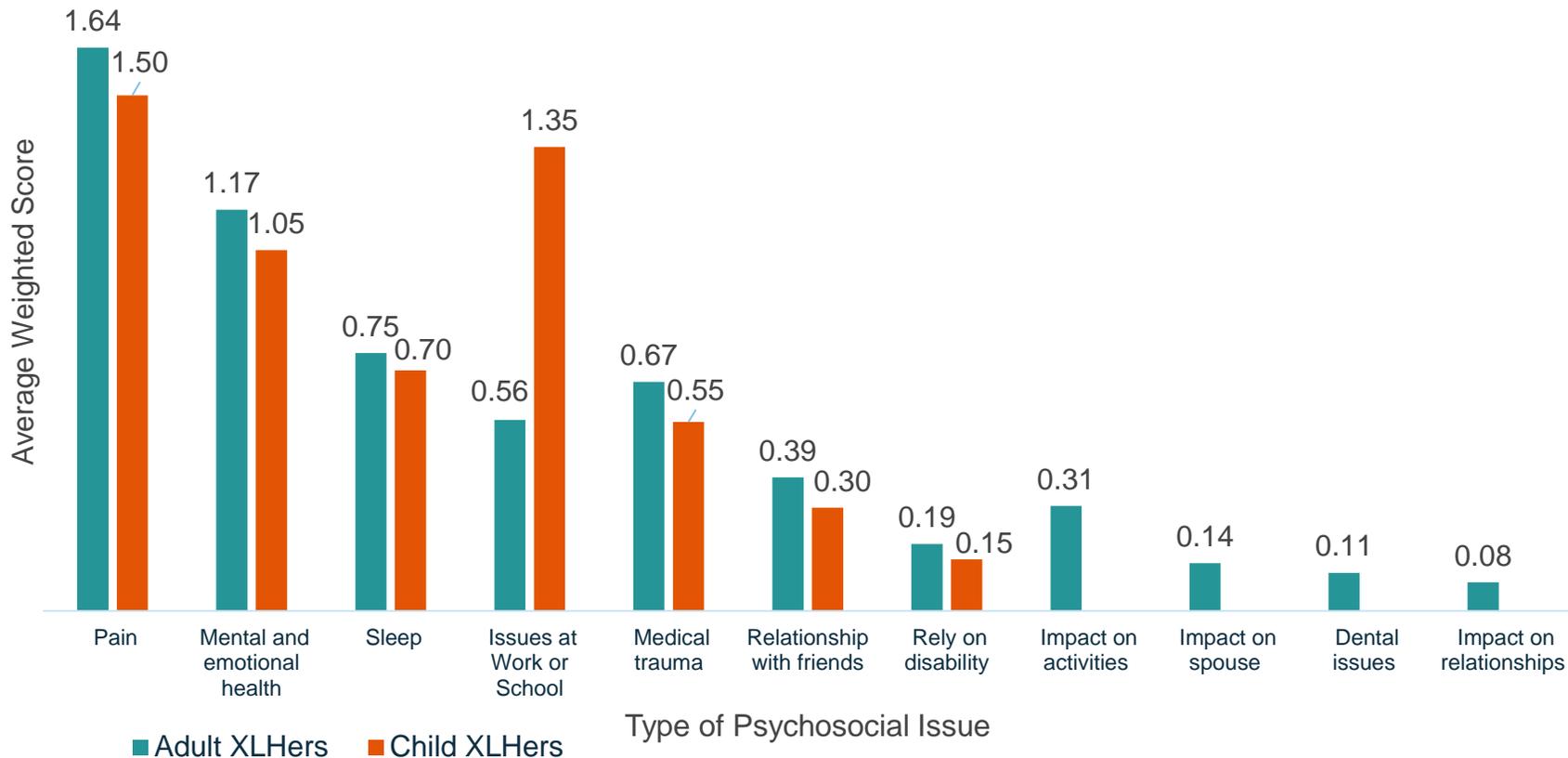
- Sertraline (3 mentions)
- Escitalopram oxalate (3 mentions)
- Fluoxetine (2 mentions)
- Amitriptyline hydrochloride (2 mentions)
- Lamotrigine (2 mentions)
- Aripiprazole (1 mention)
- Citalopram (1 mention)
- Paroxetine (1 mention)
- Bupropion hydrochloride (1 mention)
- Nortriptyline (1 mention)

Percentage of Adult XLHers Prescribed Medication for Depression



# When asked about the top 3 most burdensome psychosocial impacts, pain scored the highest for both groups; second most important impact was mental and emotional health for adult XLHers and impact on school reported by caregivers of child XLHers

Average Weighted Score – Most Burdensome Psychosocial Impacts



## Methodology for Analysis:

- In the interview, participants were asked to provide their top 3 burdens
- The #1 most important was assigned three points, the second assigned two, and the third assigned one point
- This means each respondent had a possible 6 points to assign – please note that some respondents did not have three issues to list (e.g. caregivers with small children)
- Total points were divided by the number of respondents in that category (Adults = 36, Children = 20): giving an average weighted score for each category

# Further descriptions of mental health issues

## Descriptions of Mental Health Issues Reported by XLHers (Adult and Child)

### Depression

- 10 mentions of “it comes and goes”
- 4 mentions of seasonal depression
- 3 mentions of depression due to worrying about the future
- 2 mentions each of depression due to not being able to work, being in pain, and generally lacking independence

### Social isolation

- 9 mentions of limited activity level as the cause
- 4 mentions of social anxiety as the cause
- 2 mentions of hearing loss as the cause

### Suicidal thoughts

- 5 mentions of suicidal thoughts “in the past”
- 2 mentions of “suicidal ideation”
- 2 mentions of a previous attempt

# During interviews, self-esteem, social participation, and disrupted plans were most often mentioned when discussing mental and emotional health impacts; the most common mental health symptoms mentioned were depression and anxiety

## Psychosocial Impacts of XLH - Descriptions of Mental and Emotional Health with Example Quotes

### Mental and emotional health impacts

#### Impacts on life

- Impact on self esteem when younger (10 mentions)
- Impact of social participation (5 mentions)
- Disrupted plans (4 mentions)
- 1 mention each of:
  - Fear of physical activity
  - Stress of impacting medication
  - Missing school when younger
  - Impact on sibling relationships
  - Impact on travel
  - Impact on play
  - Impact on trust in friendships

#### Mental health symptoms

- Depression (5 mentions)
- Anxiety with medical appointment/procedures (3 mentions)
- Defeat from unknowledgeable care (2 mentions)
- Social isolation (2 mentions)
- 1 mention each of:
  - Emotional toll
  - Frustration
  - Health anxiety

**“Impact on self-esteem:** *The big thing is not being able to do what his friends do, and he is shorter so that is difficult. He gets made fun of and the other kids ask questions, which has taken a toll.*

– Caregiver of child XLHer

**Impact on social participation:** *Now that my daughter is 6, she's really noticing her friends who can do a cartwheel while she can't. When we went to her doctor over the summer, we were saying how she plays softball and he said that's okay, but to stay away from soccer. There are things she's realized she shouldn't or can't do.*

– Caregiver of child XLHer

**Depression:** *There are days I wish I was normal and could get up and do what I want without XLH. It is depressing when you step out of bed and have pain. Like last Thursday, I couldn't walk or bear weight and had to call off work and I had plans that got completely shredded. I was very upset all day that day.”*

– Adult XLHer

## Quotes from adult XLHers on the impact on mental and emotional health highlight the frustration and defeat of living with a chronic condition

“

*Across my lifespan, the impact has varied. As a kid and through high school, it was about **looking different**, not hearing as well, and awkward “peer stuff”. As I got older (in my 20s), everyone was getting married, and I realized it was going to be different for me. XLH didn't have an impact on my career, but as I'm getting older, it's more challenging as my **mobility and restrictions** have increased. The hearing issues have the biggest social impact. When people are isolated, it is hard, and I can't join groups or meetups because it's hard to hear.*

– Adult XLHer

*It is discouraging to want to do something, and you **simply can't**. If you do, you are in pain, so you don't enjoy what you want to do. This time of year, I want to go shopping, but you go and can't find a close parking spot and worry about your activity making the pain worse the next day. It **wears on your mind and mental health**.*

– Adult XLHer

*Through the years growing up we were told we would “outgrow” XLH. Growing up, I would have severe bone pain in my legs, my dad would take me to the doctor, and they blamed it on growing pains. In my 20s, I started having abscesses and the dentist said it's not related to rickets. The lack of knowledge is very **frustrating and defeating**.*

– Adult XLHer”

## Quotes from caregivers of child XLHers on the impact on mental and emotional health highlight the challenges of not being able to participate in activities and looking different than other children

“

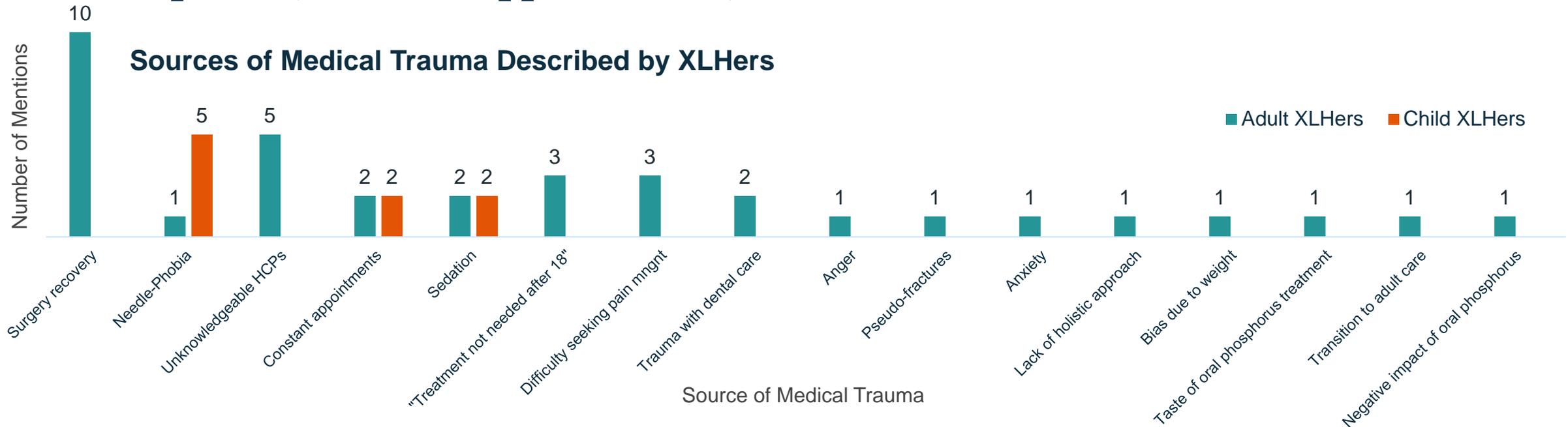
*“Kids can be mean, and this is her first year at preschool. She's never had a lot of interaction with kids not related to her. Being around people her age has highlighted that she is smaller and slower; her legs can't keep up with kids her age. Some kids don't care, and **it can make her a little sad**. She will go off and play by herself sometimes.”*

– Caregiver of child XLHer

*“As a small child, she wore braces like you would see in the movie Forrest Gump and, visibly, she was different, so she became more self-aware as she aged. We decided to do her first double knee surgery so she wouldn't have to wear the braces, which led to scars and legs still being different shaped. When she would sit crisscross, the scars would show, and others would ask her about. It made her feel **self-conscious**. One summer she refused to wear shorts because of the scars on her knees.”*

– Caregiver of child XLHer”

# When prompted to discuss medical trauma, adult XLHers most frequently mentioned recovery from surgery, whereas caregivers of child XLHers shared needle phobia, constant appointments, and sedation



*Being that my condition is so rare, the places I had surgery were not in my state, so **as a child, my parents basically left me at the out-of-state hospital** because my dad had to work, and my mom had to be with my siblings. **People have said I am outspoken, but I had to be. My parents would come to the hospital when they could, but I couldn't expect them to come every single day.** That has tremendously impacted me as an adult.*

– Adult XLHer

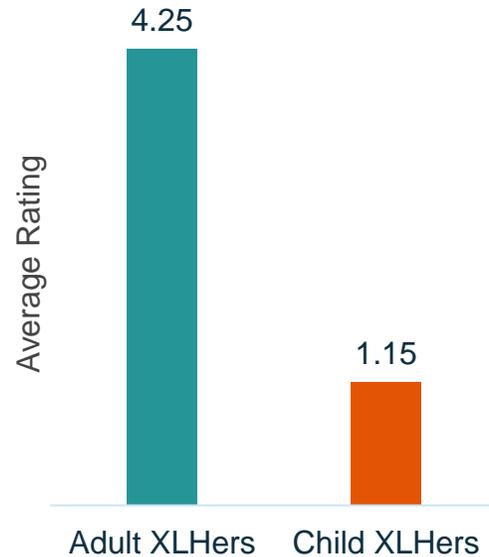
*“Her trauma is related to having to get blood drawn, and shots; it’s hard to explain all of that to a toddler... “normal” toddlers wouldn’t have to get blood drawn every few months. When first getting diagnosed, she got blood drawn 3 times in a 2-week period. Now **she sees a needle or hears that we are going to the doctor, and she cries.**”*

– Caregiver of child XLHer

n=26, 19 adult and 7 child XLHers; Most respondents (75%) rated XLH-related medical trauma on a scale of 1-10 with 10 being the most severe; if rated >3, respondents were asked more during interview about the trauma QX #42: If medical trauma >3, If < 3, skip. I want to ask specifically about medical tell me what has been traumatic about your/your child's XLH journey. You rated that number "" on a scale of 1-10, tell me what has been traumatic about your/your child's XLH journey

# Adult XLHers rated the impact of XLH on daily activities higher than caregivers of child XLHers, with the most common difficulty involving putting on socks, getting dressed, and housework

## Average Impact of XLH on Daily Activities Reported by XLHers and Descriptions



Rating of impact of XLH on daily activities on a Scale of 1-10  
1=No impact at all, 10= Extreme Impact

### Daily Activities Impacted

- Putting on socks (9 mentions)
- Getting dressed in general (7 mentions)
- Housework (6 mentions)
- Tying shoes (4 mentions)
- Driving (4 mentions)
- Mobility limitations when shopping (3 mentions)
- Cooking (3 mentions)
- Mobility limitations around the house (2 mentions)
- Ability to reach things (2 mentions)
- Showering (2 mentions)
- 1 mention each:
  - Limit activities in general
  - Laundry
  - Carrying groceries
  - Picking things up from floor
  - Carrying child
  - Gardening

5 mentions of difficulty only during surgery recovery

4 mentions of general impact on energy level

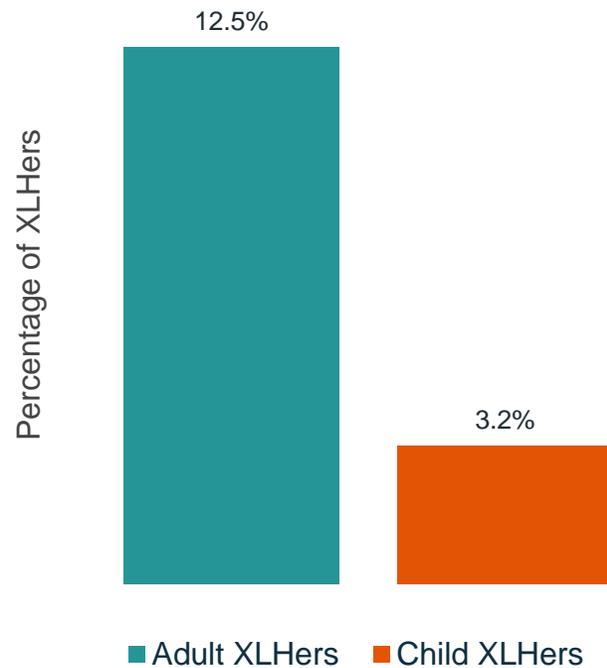
2 mentions of hearing impacting conversation

n=56, 36 adult XLHers and 20 child XLHers

QX #46: How much would you say these issues or other XLH-related burdens impact your / your child's ability to care for yourself/ him or herself or your family on a scale of 1-10 with 1 being no impact at all and 10 being extreme impact? (i.e. getting dressed, cooking, doing chores, driving, etc.) Please describe why you answered like you did

# While the study did not probe specifically about how psychosocial issues are managed, 7 adult XLHers and 1 child XLHer currently see a mental health professional

**% of XLHers who see a “Psychology, Psychiatry or Mental Health” Professional**



Average OOP cost for mental health care	Range
\$605	\$120-\$2,000

### Items Noted as Mental Health Out-of-Pocket Costs

- Therapist (4 mentions)
- Copay/Deductibles (3 mentions)
- Medications (2 mentions)
- Initial Consult (1 mention)
- Upgrading Insurance Policy (1 mention)
- “Manic depressive” (1 mention)

N=87, 56 adult XLHers and 31 child XLHers

QX# 25 and 11: In the last year, have you paid any of the healthcare costs associated with XLH out of pocket? Please estimate the amount that was spent (on mental health care). Describe what this was spent on.

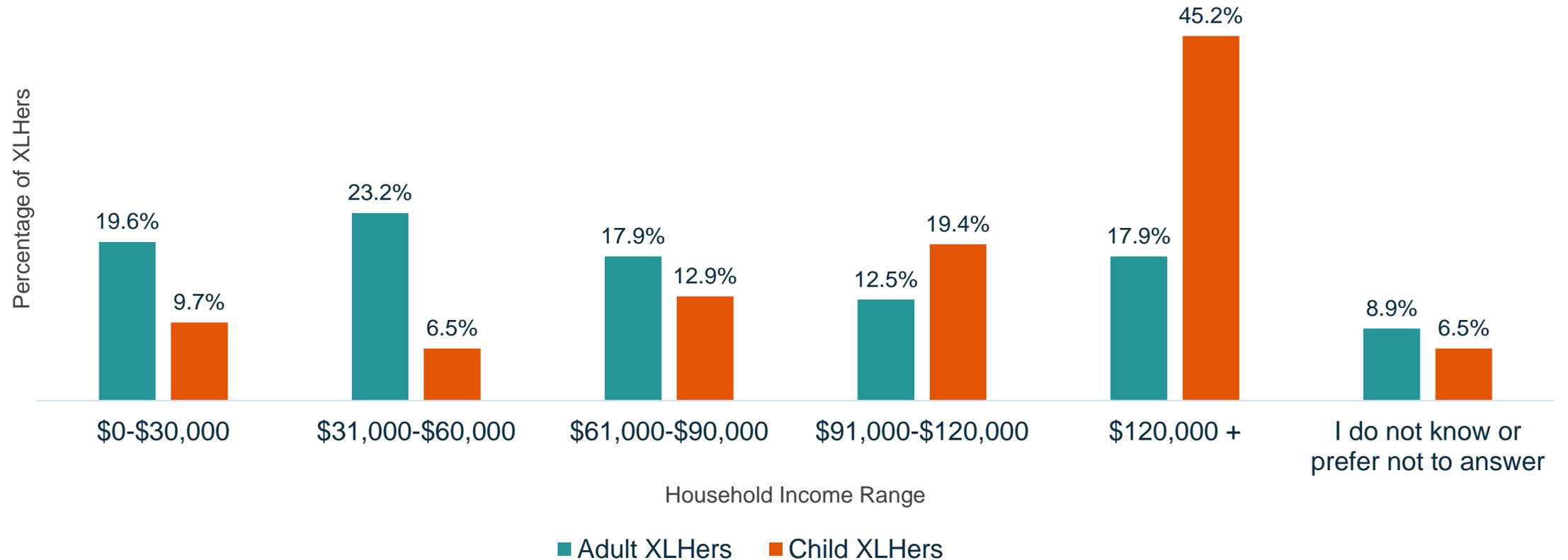
Please select all options that describe the specialty of the healthcare providers who you/your child currently see for various issues related to XLH. (Please note, the healthcare provider does not have to be a doctor) \*Psychology, psychiatry or mental health was an option in the list

# Financial Burden

- Household Income
- Health & Dental Insurance Coverage
- Assistance Programs
- Out-of-pocket Costs
- Indirect Financial Impacts

# Household income for adult XLHers was fairly evenly distributed, though more caregivers of child XLHers fell into the \$120k+ category

## Annual Household Income of Survey Respondents

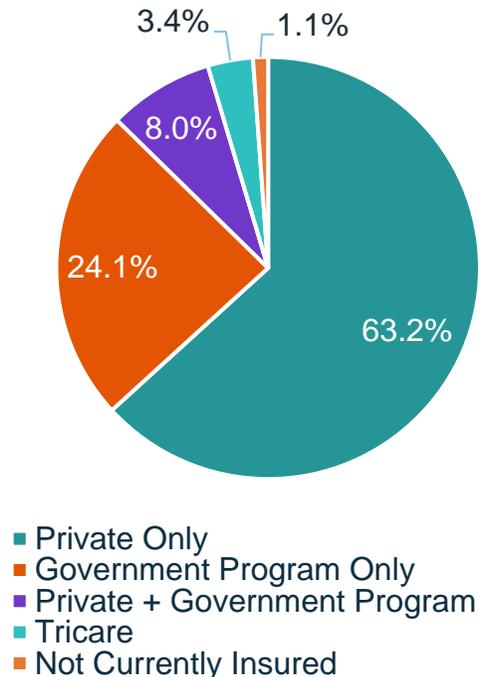


# Most XLHers (63.2%) were covered by private insurance only, followed by government provided insurance (24.1%); only 1 participant was not currently insured

## Insurance Coverage Breakdown

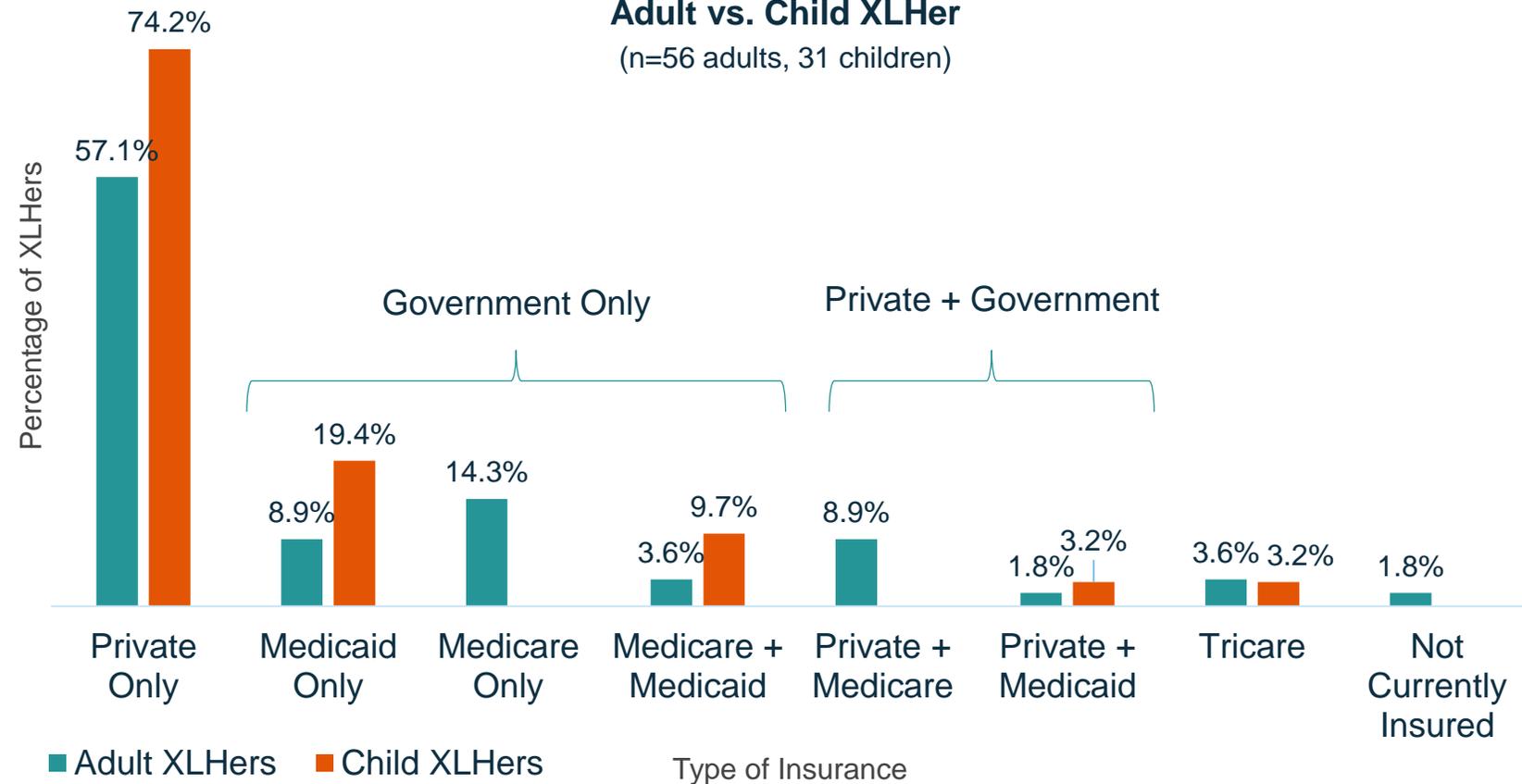
### All Survey Respondents

(N=87)



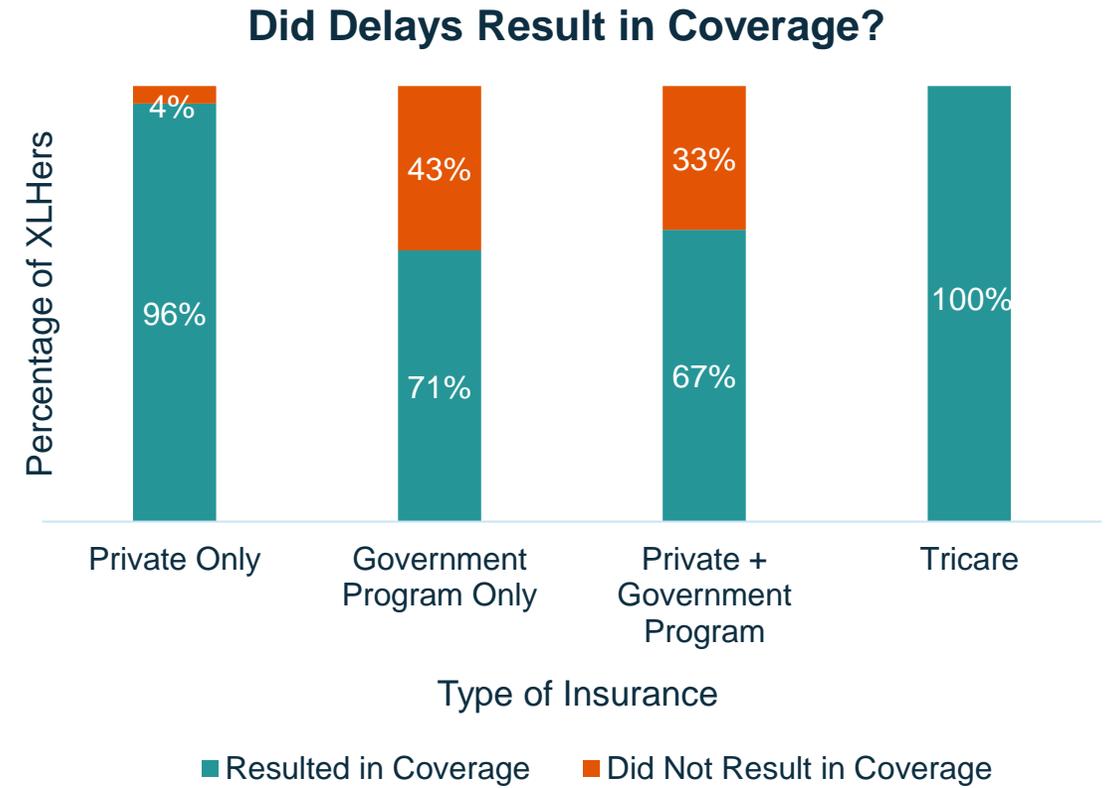
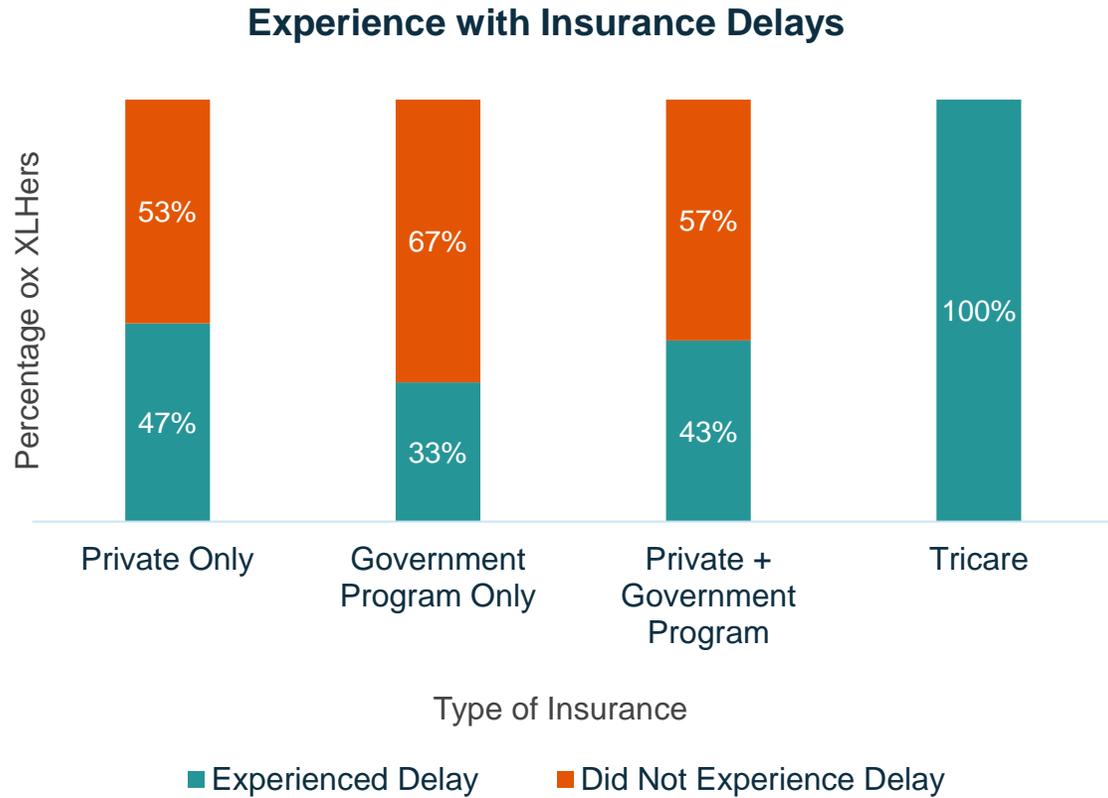
### Adult vs. Child XLHer

(n=56 adults, 31 children)

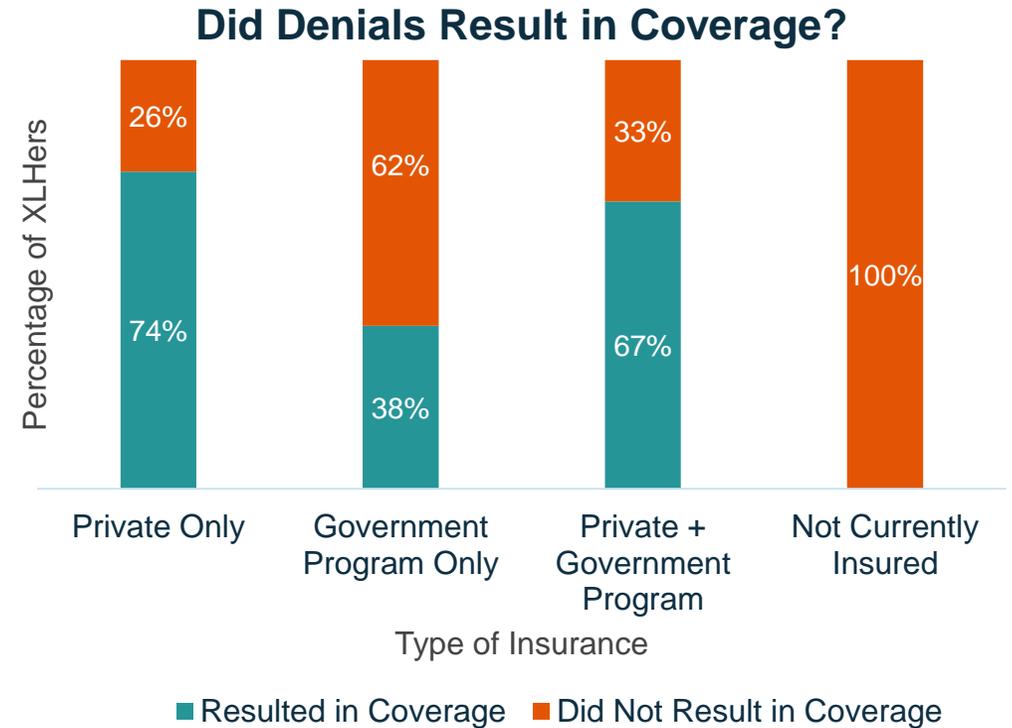
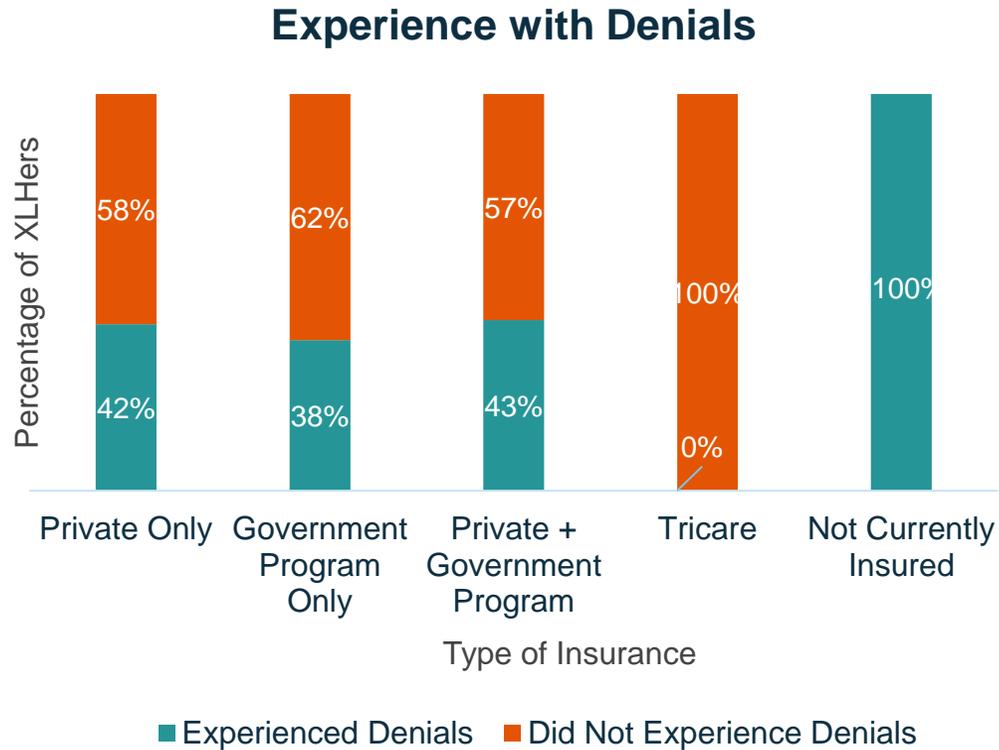


QX #18: Please select the options that best describe the type of healthcare insurance that covers you/your child (the person with XLH who is the subject of this survey)  
 QX #57: Next I want to talk about the financial impact that XLH has had on you or your family. I understand that for health insurance you are currently covered by <insurance type> - can you provide the name of your insurance company?

# Respondents in each of the insurance groups experienced delays in coverage, though XLHers with "government program only" cited fewer delays; the majority of delays for all respondents resulted in coverage, rather than denials

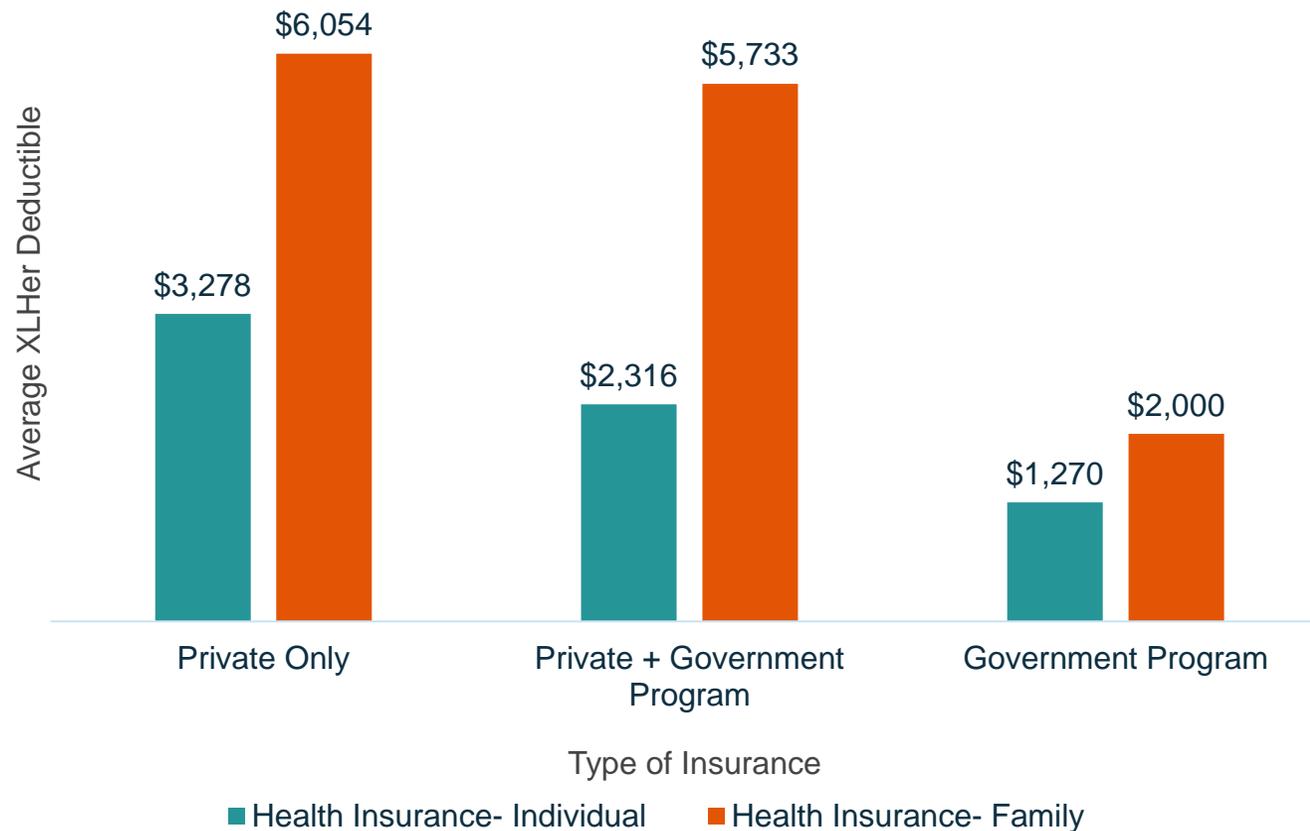


**Respondents in each of the insurance groups (other than Tricare) experienced coverage denials; for those with private or private + government programs, denials often resulted in coverage whereas those who are uninsured or on “government programs only” typically did not**



# When in place, insurance deductibles were up to \$8,000 for individuals and \$13,500 for families; those on government-only plans were most likely to say there was no deductible

Average Deductible, by Insurance Group of XLHers and Descriptions



### Government Program

- 9 noted no individual deductible, 17 noted no family deductible
- Range: \$0-8,000 for both individual and family

### Private + Government Program

- 1 noted no individual deductible and no family deductible
- Range: \$300-6,000 for individual and \$500-10,000 for family

### Private Only

- 19 noted no individual deductible, 28 noted no family deductible
- Range: \$450-8,000 for individual and \$900-13,500 for family

### Tricare

- 1 noted \$300 individual and \$3,000 family
- 2 noted no individual or family deductible

### Not Currently Insured

- Question is not applicable

# Most (81.6%) XLHers in the study reported having dental insurance; those with private insurance or private insurance + government were most likely to have dental insurance

Percentage of XLHers with Dental Insurance, by Insurance Type

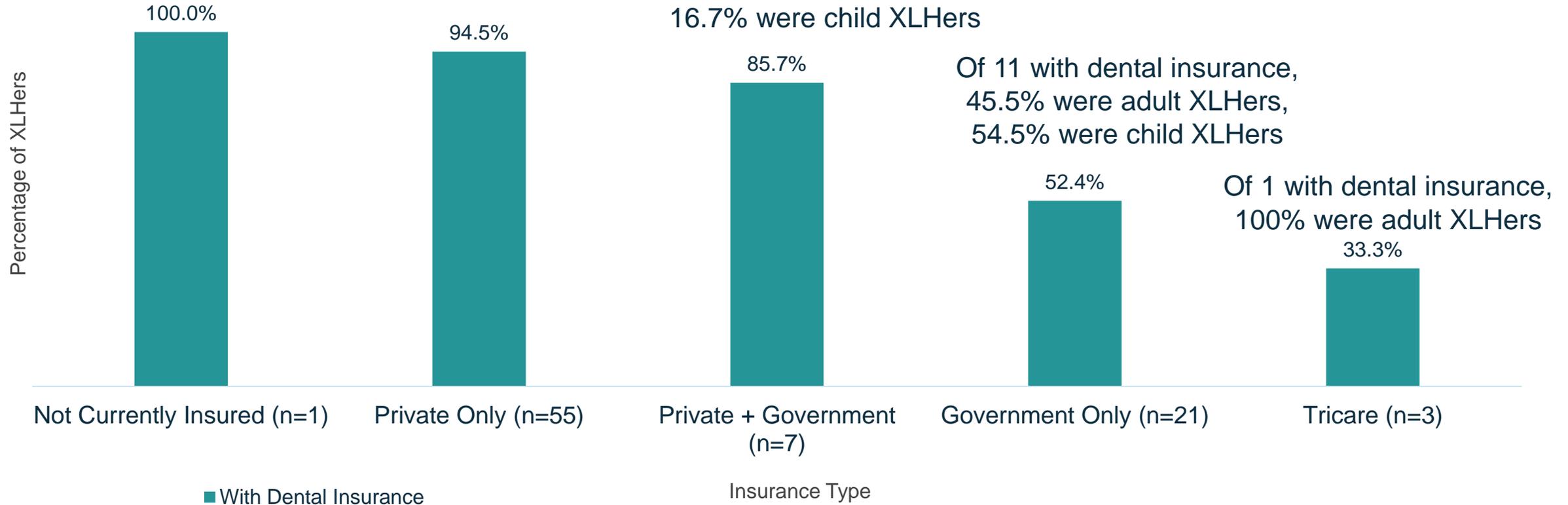
Note: this respondent noted having no health insurance but having dental insurance, situation unclear

Of 52 with dental insurance, 57.7% were adult XLHers, 42.3% were child XLHers

Of 6 with dental insurance, 83.3% were adult XLHers, 16.7% were child XLHers

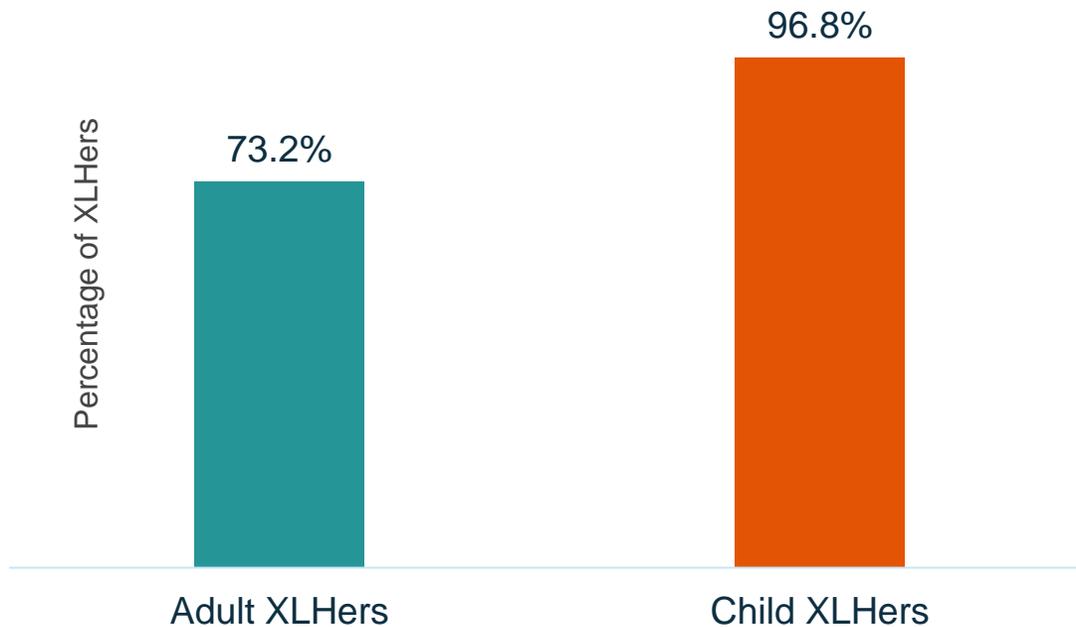
Of 11 with dental insurance, 45.5% were adult XLHers, 54.5% were child XLHers

Of 1 with dental insurance, 100% were adult XLHers



# Of the 71 (81.6%) XLHers reported to have dental insurance, 41 were adult XLHers and 30 were child XLHers; Delta Dental was the most frequently mentioned plan

### Comparison of Adult vs Child XLHer Dental Insurance Coverage

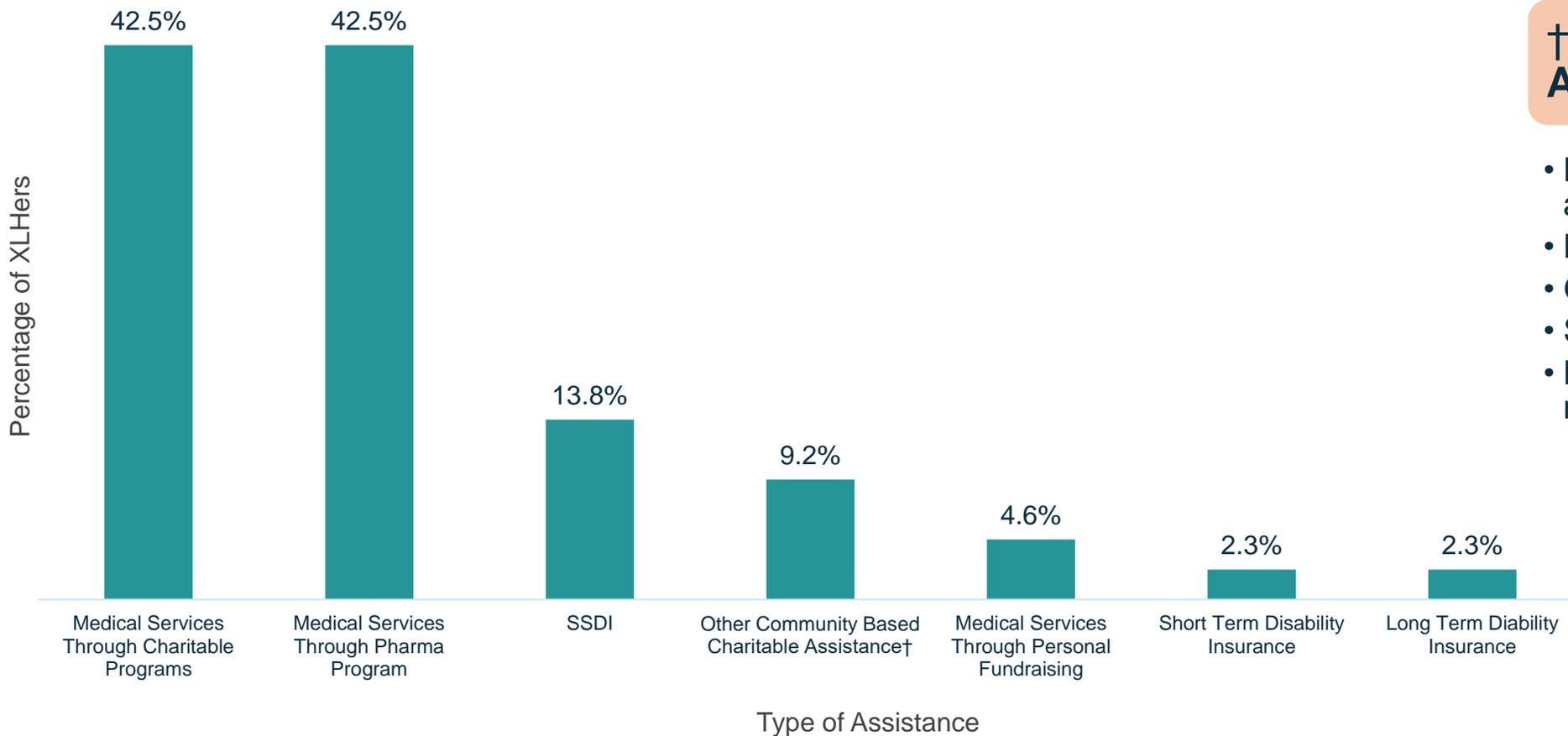


### Dental Insurance Companies, Captured During Interviews (44 mentions)

- Delta (11 mentions)
- Cigna (5 mentions)
- Guardian (5 mentions)
- United (4 mentions)
- Medicaid (3 mentions)
- BCBS (3 mentions)
- Metlife (3 mentions)
- Medicare (2 mentions)
- 1 mention each:
  - Aetna
  - AFLAC
  - Atlas
  - Cobra
  - Dental select
  - Denti-Cal
  - Dominion
  - GIHA

# Primary forms of assistance used by survey respondents include Charitable Programs (e.g., Assistance Fund) and Pharma-Sponsored Programs; no notable differences between age groups

Forms of Assistance Accessed by Survey Respondents\*



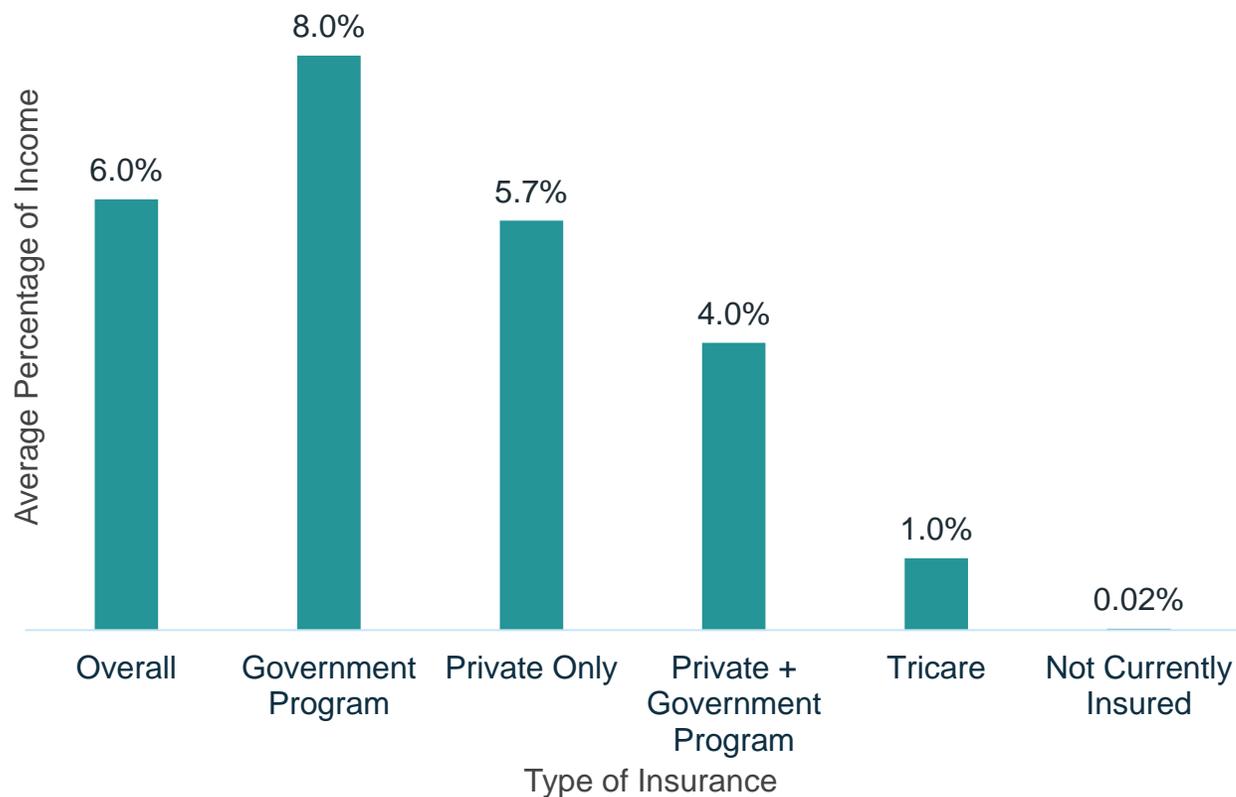
**†Other Forms of Assistance Noted:**

- Health reimbursement arrangement through work
- Home health care
- Grant through doctor’s office
- School lunch program
- Noted none currently but “will need soon”

\*Some respondents used assistance from multiple sources

# The average percentage of income that went to out-of-pocket costs in the last year was highest for those on government programs, followed by those with private insurance plans

**% of Annual Income Spent on Out-of-Pocket Costs in the Last Year**



Type of Insurance	Range of Annual Income % Spent on OOP Costs
Overall	0-40%
Government Program	0-25%
Private Only	0-40%
Private + Government Program	0-10%
Tricare	0-3%
Not Currently Insured	0.02%

# Average out-of-pocket costs in the past year were highest for home modifications for accessibility, dental care, and assistive devices / medical equipment

## Out-of-Pocket Expenses in Past Year

Expense Category	Average OOP	OOP Range	Top 2 Most Frequently Mentioned Expenses (# mentions)
Home Modifications (n=9)	\$6,919	\$52-\$40,000	Ramp/wheelchair access (3)
Dental (n=51)	\$2,775	\$30-\$40,000	Extractions (9) Crowns (9)
Other (n=12)	\$2,024	\$32-\$8,300	(see single mentions on next slide)
In-Network OOP Costs (n=42)	\$1,304	\$50-\$12,000	Copay/deductible (8) Office visits (6)
Out-of-Network Providers (n=13)	\$1,263	\$100-\$5,000	(see single mentions on next slide)
Travel (n=49)	\$1,002	\$10-\$15,000	Fuel (20) Lodging (7)
Physical Therapy (n=19)	\$788	\$31-3500	Copay/deductibles (6) Unapproved PT (5)
Mental Health Care (n=12)	\$605	\$120-\$2,000	Therapist (4) Copay/deductibles (3)
Pain Management (n=26)	\$382	\$10-\$3,300	Over-the-counter (OTC) medications (9) Copay/deductibles (5)

- Most (60.9%) XLHers in the study **paid some dental expenses out-of-pocket in the last year**
- **Among XLHers with OOP expenses over \$1000, 72.7% reported having dental insurance** suggesting that current dental insurance plan do not meet their needs

# Out-of-pocket expenses reported by survey respondents in the past year

(# mentions of each expense, by category)

Home Modifications	Dental	Other	In-Network OOP Costs
Ramp/wheelchair access (3)	Extraction (9)	Walker	Copays/Deductible (8)
Driveway	Crowns (9)	Scooter	Office visits (6)
Bathtub	Root canal (6)	Scooter batteries	Lab work (4)
Lower toilet/mobile toilet seat	Cleaning (6)	Shoe lift	X-rays (2)
Bike modifications	Implants (3)	Socks	Travel
Handles/hardware	Consults/check-up (3)	Reaching device	Cortisone injections
Shower seat	Orthodontics/braces (3)	Bath sponge devices	Cast for broken foot
	Dentures/partial dental plate (2)	Medical equipment	"Assorted Care"
	Travel to dental services (2)	Lost wages	"Specialist – Endocrinologist"
	Filling (2)	"Food/hotels too far"	"Local endocrinology, children, pulm"
	Copay (2)	"Magnesium flakes for help with restless legs"	"Renal US"
	3D x-ray	"Magnesium rubs to help with restless legs"	"Military doctor"
	Uncovered services	"Over counter pain meds like Tylenol, ibuprofen "	
	Dental issues unspecified	"Pharmacy co-pay"	
	Sealants	"Orsini"	
	Pallet expander		
	Sedation		
	Cap		
	Maxillofacial Surgery		

# Out-of-pocket expenses reported by survey respondents in the past year (continued)

(# mentions of each expense, by category)

Out-of-Network Providers	Travel	Physical Therapy	Mental Health Care	Pain Management
"Manic depression"	Fuel (20)	Copay/Deductibles (6)	Therapist (4)	Over-the-counter medications (9)
"XLH endocrinologist"	Lodging (7)	Unapproved PT (5)	Copay/Deductibles (3)	Copay/Deductibles (5)
"Visit payments"	"Travel/Travel costs" (3)	Knee (2)	Medications (2)	Unapproved PT (4)
"Co-pay"	Parking (2)	Shoulder	Initial Consult	Acupuncture (2)
"Yale yearly visit"	Tolls	PT Equipment	Upgrading Insurance Policy	Prescription medication (2)
	Rental vehicle	Private Trainer	"Manic depressive"	Massage therapy/OMT (2)
	"Trips to dental school, other specialists"	Travel		Electric motorcycle/heating pad
	"Appointments"			Uncovered visit
	Airfare			Travel
	Tires/Car Services			Pain pump
	Time			"Only post-surgery"
	"Psychiatrist"			
	"Travel costs 1.75 hrs each way 100 miles"			
	"Having to travel to endocrine"			

# Interviewees cited the following indirect financial impacts of XLH: missing work, having to use PTO or medical leave, and gas used to get to and from appointments

## Indirect financial impacts of XLH

- 21 mentions of missing work
- 5 mentions of using PTO
- 3 mentions each of medical leave and gas
- 2 mentions each of having to leave a job (retirement), Ubers, food when travelling to appointments, supplements, getting clothes tailored
- 1 mention each of handles for the bathtub, 'grabber tool,' shoehorn, special sports bras, sponge for shower, childcare, lidocaine patches, ibuprofen, compression gloves deep tissue massages, parking fees, expenses for conferences, and in-home help



*I tend to use PTO or work late to make up time. I am lucky to have paid time off even though it is not ideal to use it for visits and not vacation. I also keep a lot of supplies, like I always have lidocaine patches, ibuprofen, supplements, compression gloves, deep tissue massages... that all adds to at least \$1000, probably closer to two [thousand].”*

– Adult XLHer

*We have a nurse who comes to the house, so I schedule that when I'm not working. But there are some appointments where I drive her to the city and **take the days off**, these are typically endocrinology and orthopedics about 4 times per year.”*

– Caregiver of Child XLHer



# XLH Community Impact Survey