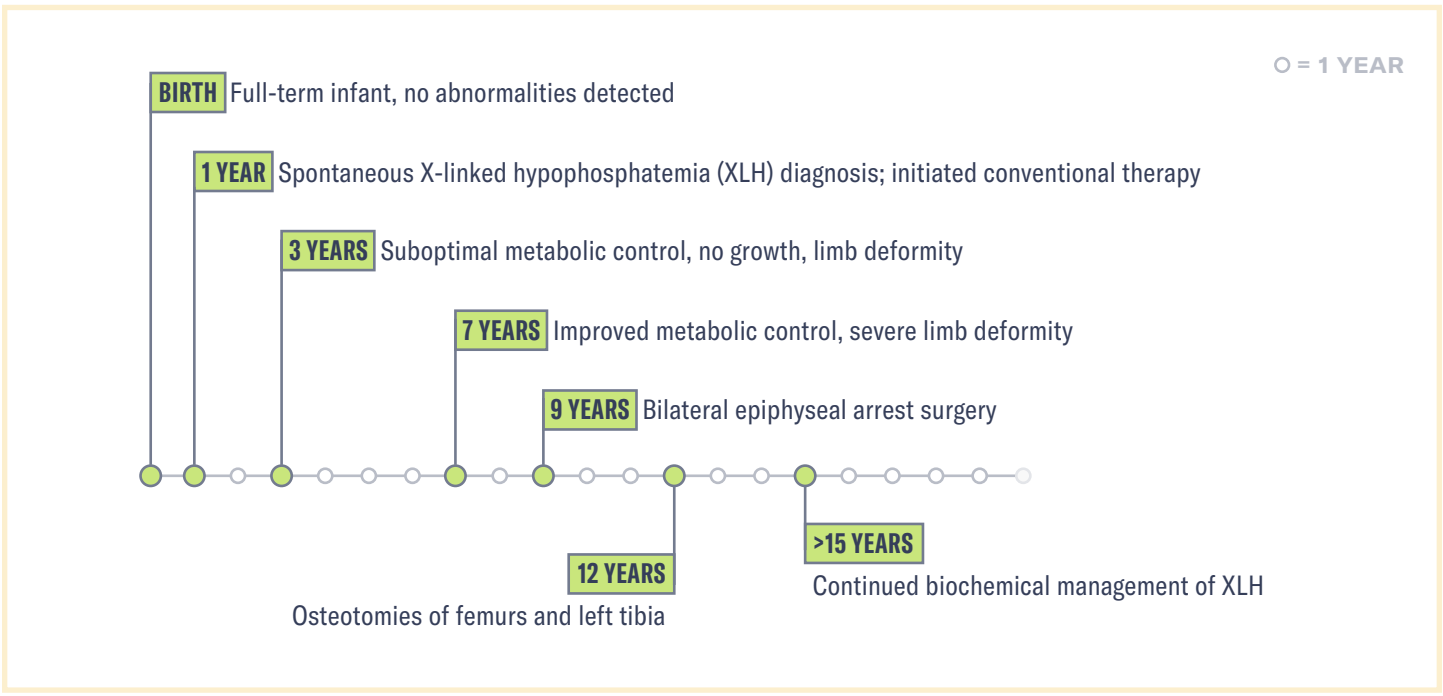
15-year-old female with spontaneous XLH*



Case summary

Findings/outcomes

- The goal of management aimed to optimize bone health before growth plate closure
- Regardless of provider, asking about mental health was an important element of care
- Once growth potential was reached, management of XLH was not over; goals shifted from promoting bone growth to fracture prevention and maximizing musculoskeletal health

Symptoms of XLH are nonspecific when considered individually and vary between patients. Specialist referral is key to early disease management.

*The information for this case study was provided courtesy of Dr. Anthony Portale, Director of the Pediatric Dialysis Program, UCSF Benioff Children's Hospital, San Francisco. This case study represents a real patient and is intended to be illustrative, not a recommendation for treatment or management. This case study does not claim to represent typical results.

Medical history

Birth

- Full-term infant with no abnormalities detected

12 months

- Signs consistent with pain upon weight bearing; limping; bowed legs at 18 months

Family history

- Unremarkable

30 months

- X-rays: bilateral bowing with irregular and widened metaphyses at tibia and femur

34 months: Key fasting laboratory findings (reference range†)

- Serum phosphorus: **2.4 mg/dL** (4.3-6.8 mg/dL)
- 25-hydroxyvitamin D: 39 ng/mL (≥20 ng/mL)
- Alkaline phosphatase: **512 U/L** (156-369 U/L)
- Serum calcium: 9.2 mg/dL (9.2-10.5 mg/dL)
- Parathyroid hormone: **85 ng/L** (10-65 ng/L)

†Indicates normal range, age and sex matched. Note that normal range values may vary depending on reference dataset. These ranges were provided by the treating physician. Colored values are outside of the normal range provided by the physician and can raise suspicion of XLH.

Growth chart: Stature for age percentiles, girls aged 2-20 years[1‡]

History of growth progression:

- Patient had short stature. Linear growth accelerated from 8 to 10 years of age, consistent with early growth spurt

‡The reference percentiles on the graph are combined from the 2 clinical growth charts for girls 2-20 years of age provided by the Centers for Disease Control and Prevention.

Diagnosis and initial treatment

1 year of age

- Spontaneous XLH
- Treatment: oral calcitriol and phosphate

Disease progression

1 to 3 years of age: Managed under orthopedic care

- Optimal control of metabolic bone disease difficult to achieve
- Patient had short stature (see Growth chart on the left)

3 years of age: Referred to endocrinology

5.9 years of age

- Musculoskeletal presentation with limb deformity in femur, tibia, and fibula; rachitic changes (see X-ray 1 on the right)

Although this patient's laboratory results improved, there was no height velocity increase. At 3 years of age, it was concluded that biochemical optimization was not met; patient was then seen monthly to tailor therapy.

7 years of age: Metabolic bone disease under control

- Surgery of lower left extremity discussed; postponement recommended
- Reported some classmates teased her for wearing a brace
- Parents reported good activity level

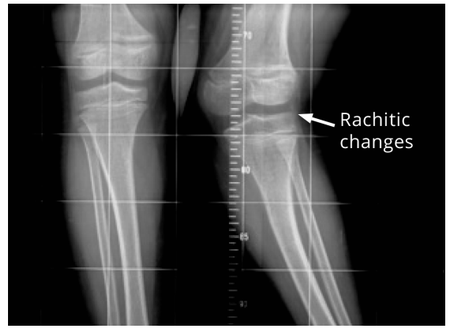
3 to 9 years of age

- Growth velocity normal (see Growth chart on the left)

9.5 years of age

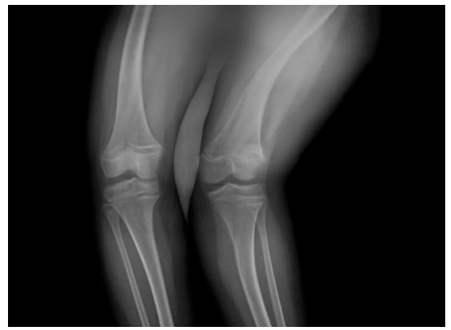
- Musculoskeletal presentation at prominent genu valgum, severe angulation at left knee with knees touching (see X-ray 2 on the right)

X-ray 1: **Limb deformity**



5.9 years of age: limb deformity: femur, tibia, and fibula; rachitic changes; lateral angulation at left knee.

X-ray 2: **Severe left knee lateral angulation**



9.5 years of age: prominent genu valgum of left lower extremity; severe lateral angulation at left knee, with knees touching; 16 cm between medial malleoli, minimal medial angulation of right lower extremity; wide-based gait.

As this patient matured, awareness of disability and obvious orthopedic deformities increased. Regardless of provider, asking about her mental health was an important element of care.

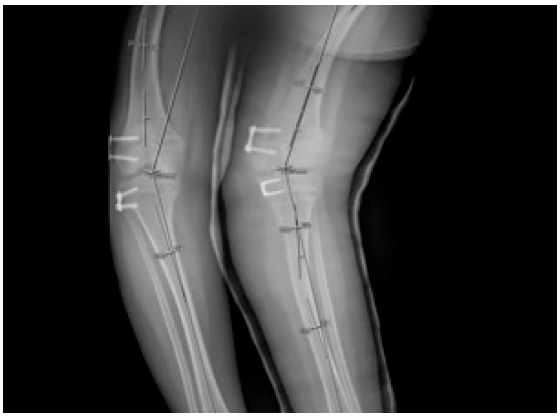
XLH LINK

Disease progression (cont'd)

10 years of age

- Linear growth accelerated consistent with early growth spurt; patient was at the 5th percentile for height (see Growth chart on inside)
- Underwent bilateral epiphyseal arrest surgery (epiphysiodesis) (see X-ray 3 on the right)
- Increasing complaints of muscle weakness and left hip, leg, and back pain
- Tired very easily and could walk only short distances without resting
- Limb deformity worsened despite treatment with oral calcitriol and phosphate
  - Exhibited signs of secondary varus of right knee as in windswept deformity

X-ray 3: **Windswept deformity**

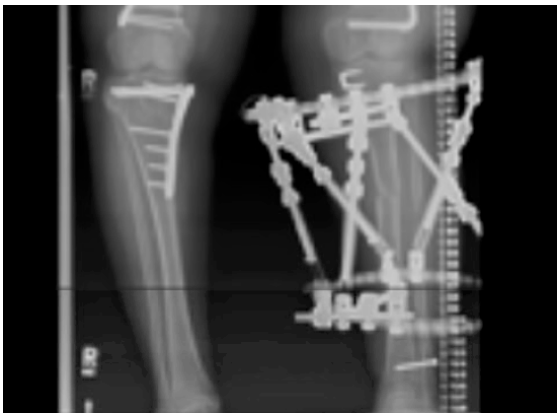


10 years of age: epiphyseal arrest surgery; windswept deformity.

Between 12.5 and 14.5 years of age

- Patient underwent successful osteotomies of femurs and left tibia (see X-ray 4 on the right)
- Homeschooled

X-ray 4: **Osteotomies**



12.5 to 14.5 years of age: osteotomies of femurs and left tibia.

Once growth potential was reached, XLH management continued, including monitoring of biochemical bone health to promote healing and peak bone mass. As the patient aged, her disease management transitioned from promoting bone growth to fracture prevention and maximizing musculoskeletal health.

REFERENCE:

1. Centers for Disease Control and Prevention. Clinical growth charts. 2 to 20 years: girls. Published May 30, 2000. Accessed May 21, 2019. https://www.cdc.gov/growthcharts/data/set2clinical/cj41c072.pdf

KYOWA KIRIN

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