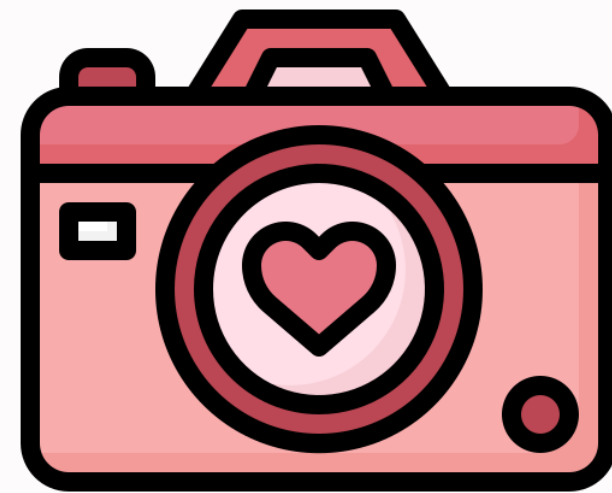


XLHuk.org

Community Day 2026

Welcome!

Housekeeping



XLHuk.org

XLHuk



Agenda for the Day

11.10: Professor Padidela

Comfort break

11.45: Dr Fiona Kirkham-Wilson

12.15: Connecting as a community

12.30: Group photo

Lunch

14.00: Networking & Fun

15.00: Close



Our Purpose

XLH UK exists to help those with XLH and related disorders within the UK, through research, support and advocacy.

Events

Submissions

News

We...

Listen

Represent

Inform

Research

Patient stories

Webinars

Hearing from the Professionals



Professor Raja Padidela



**Consultant Paediatric
Endocrinologist & Metabolic
Bone Diseases**

Royal Manchester Children's Hospital
Oxford Road, Manchester, M13 9WL

**Professor & Honorary Clinical
Chair**

Faculty of Biology, Medicine and Health,
The University of Manchester

X-Linked Hypophosphatemia

Dr Raja Padidela

Consultant Paediatric Endocrinology &
Metabolic Bone Diseases

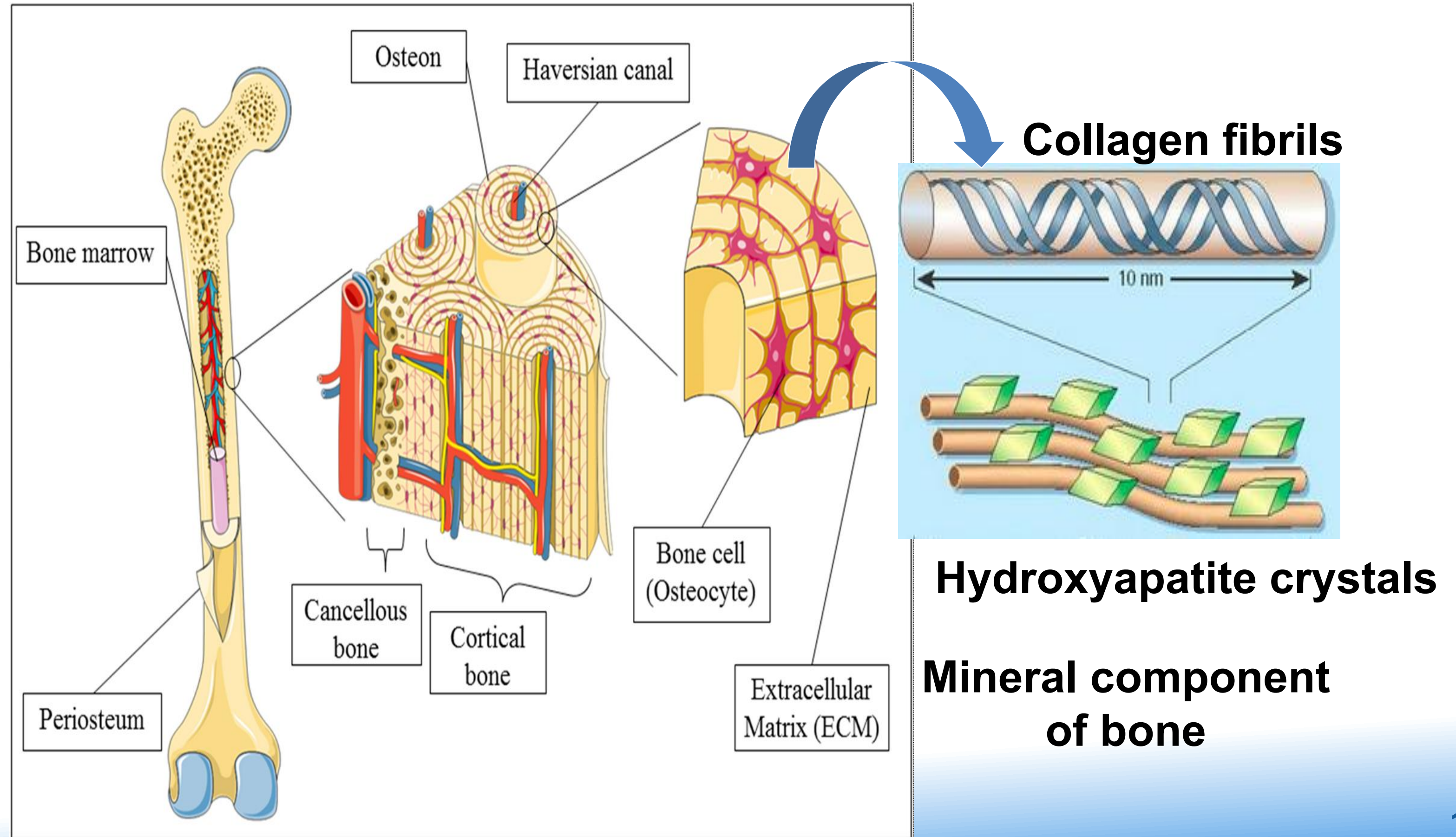
Royal Manchester Children's Hospital



Outline

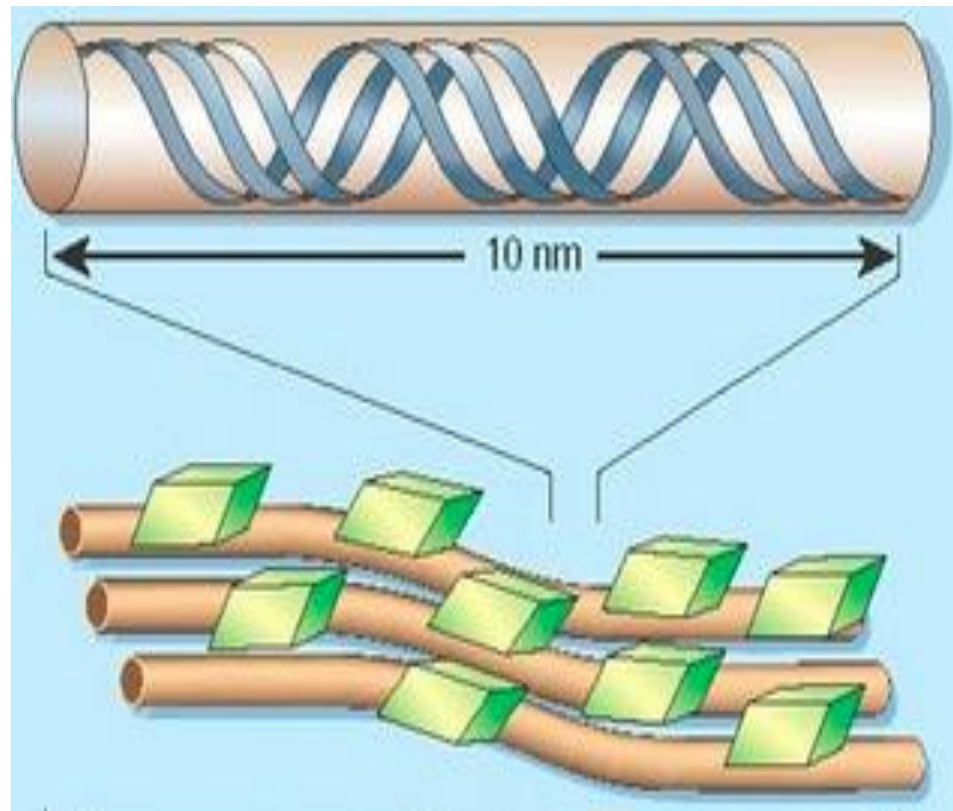
- What is X-linked hypophosphatemia (XLH)
- What causes XLH
- Limitations of previous standard of care
- Recent advances in management of XLH

Structure of bone



Structure of bone

Collagen fibrils

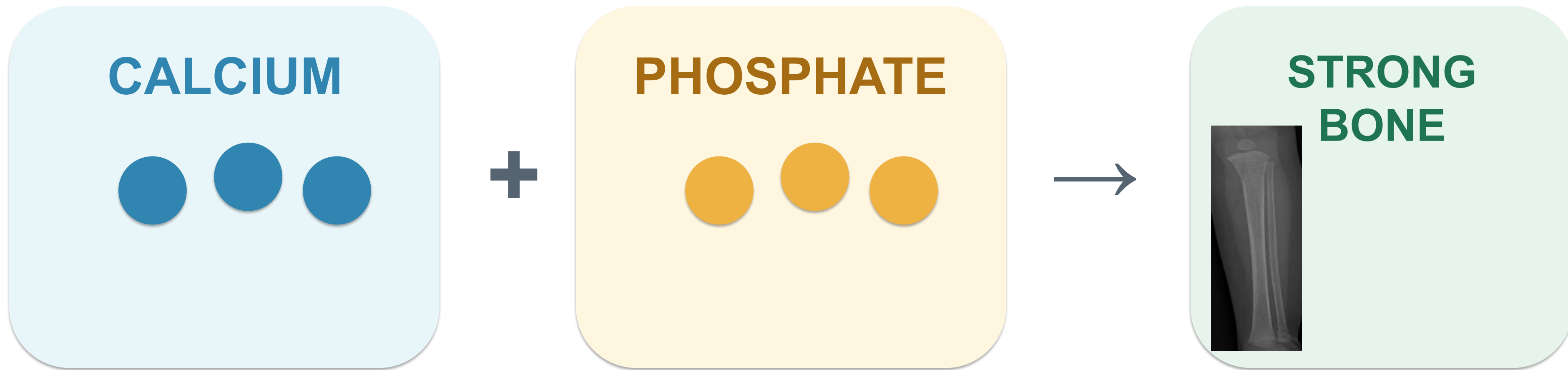


Hydroxyapatite crystals

**Mineral component
of bone**

**Calcium +
Phosphate**

What is the role of minerals in the bones?



Together they form hydroxyapatite crystals — the mineral that gives bones hardness.

Why is phosphate important during childhood?

Children need phosphate to build and mineralise growing bones.

With enough phosphate



Bones can grow straighter and stronger

With too little phosphate (XLH)



Bones may become softer or bowed

XLH causes phosphate loss through the kidneys, making normal bone mineralisation harder.

X-linked Hypophosphatemia (XLH)

- Caused by Excess FGF23
- X-linked dominant condition caused by mutation of *PHEX* gene
- Global prevalence of ~1/20,000



US



Canada



Germany



UK



France



Spain



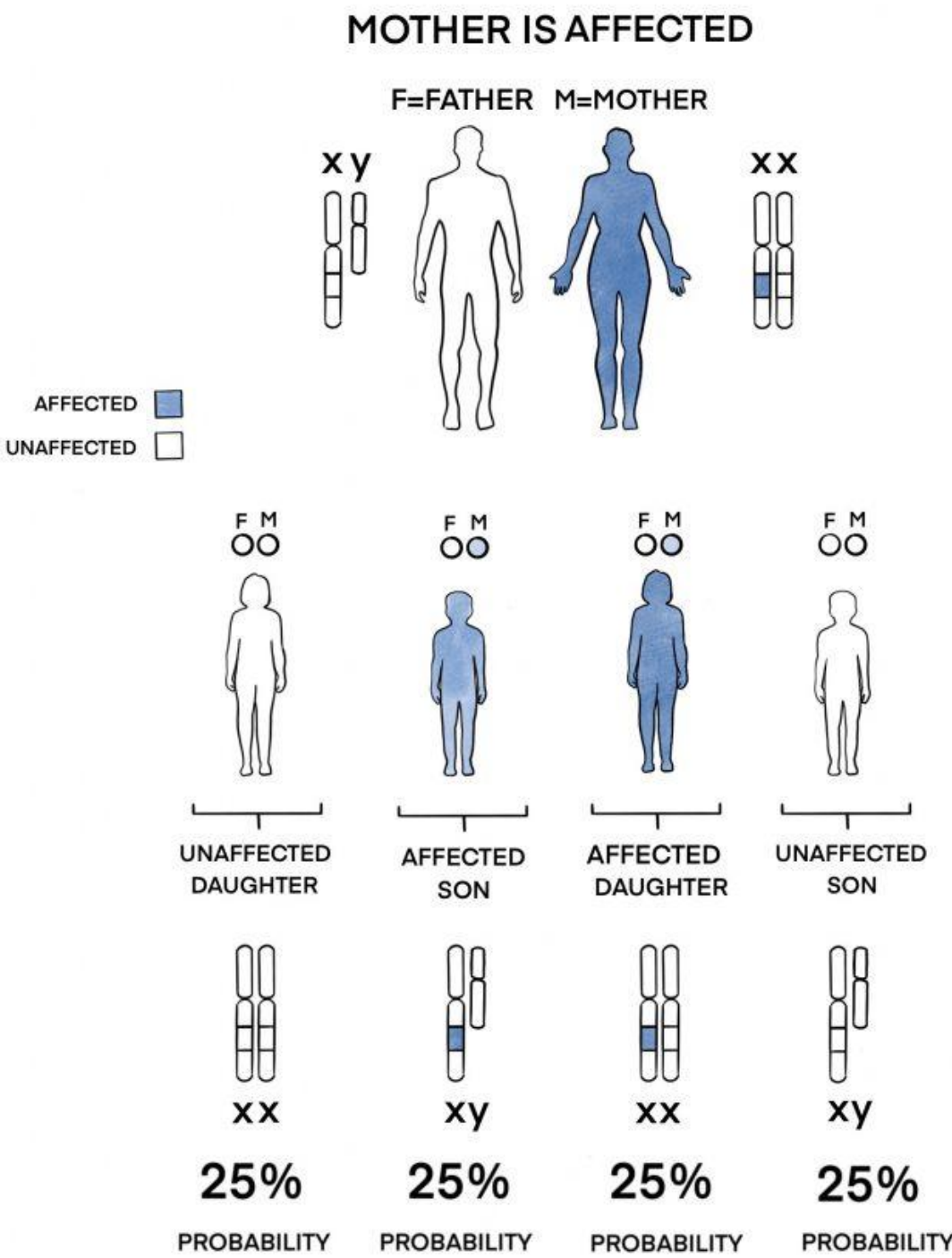
Italy

Estimated number of patients based on prevalence of XLH

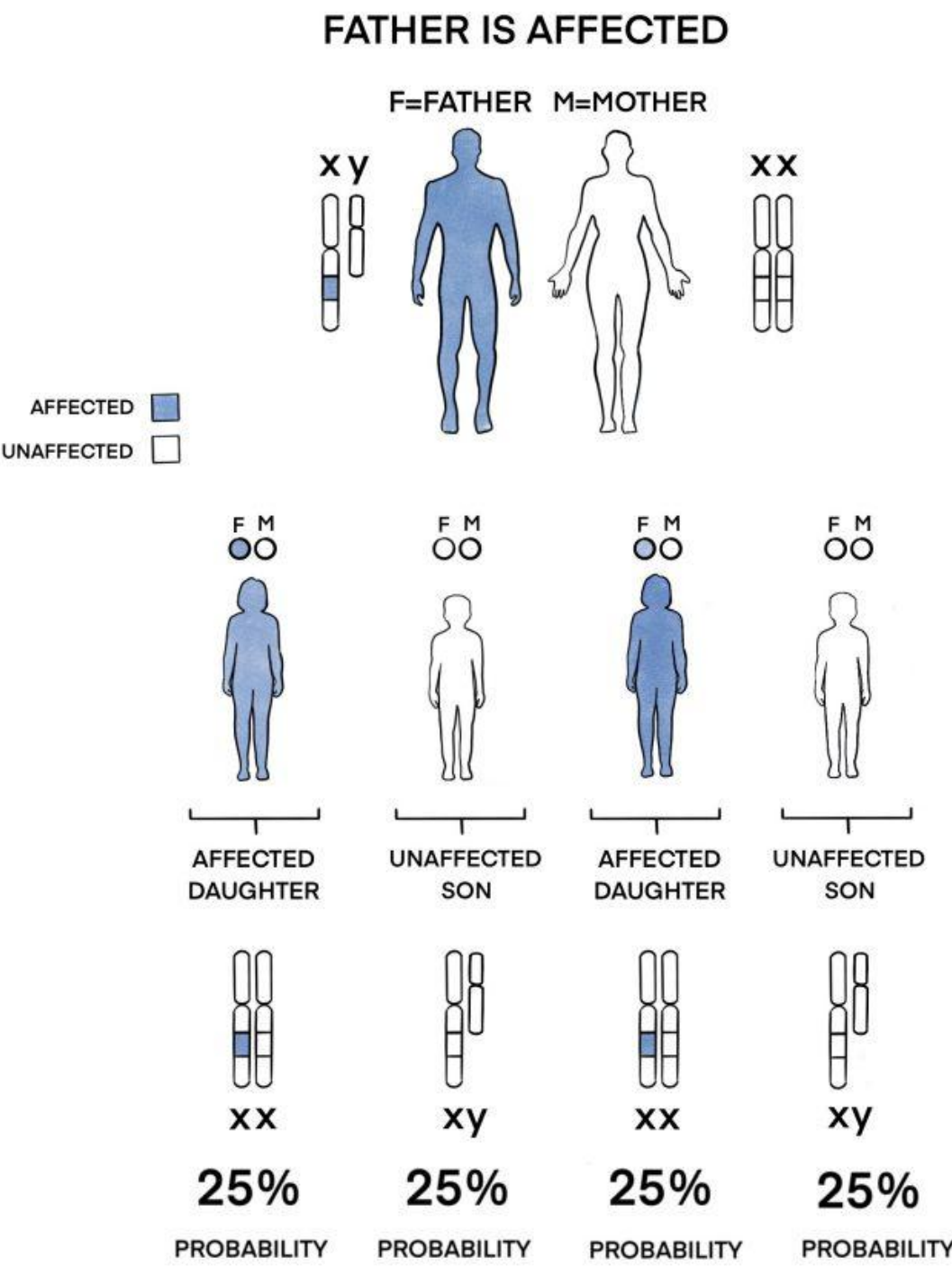
Adult	12,400	1,400	3,400	2,600	2,500	2,500	1,900
Pediatric ¹	3,700	300	600	700	700	500	400
Total	16,100	1,700	4,000	3,300	3,200	3,000	2,300

X-linked Dominant inheritance

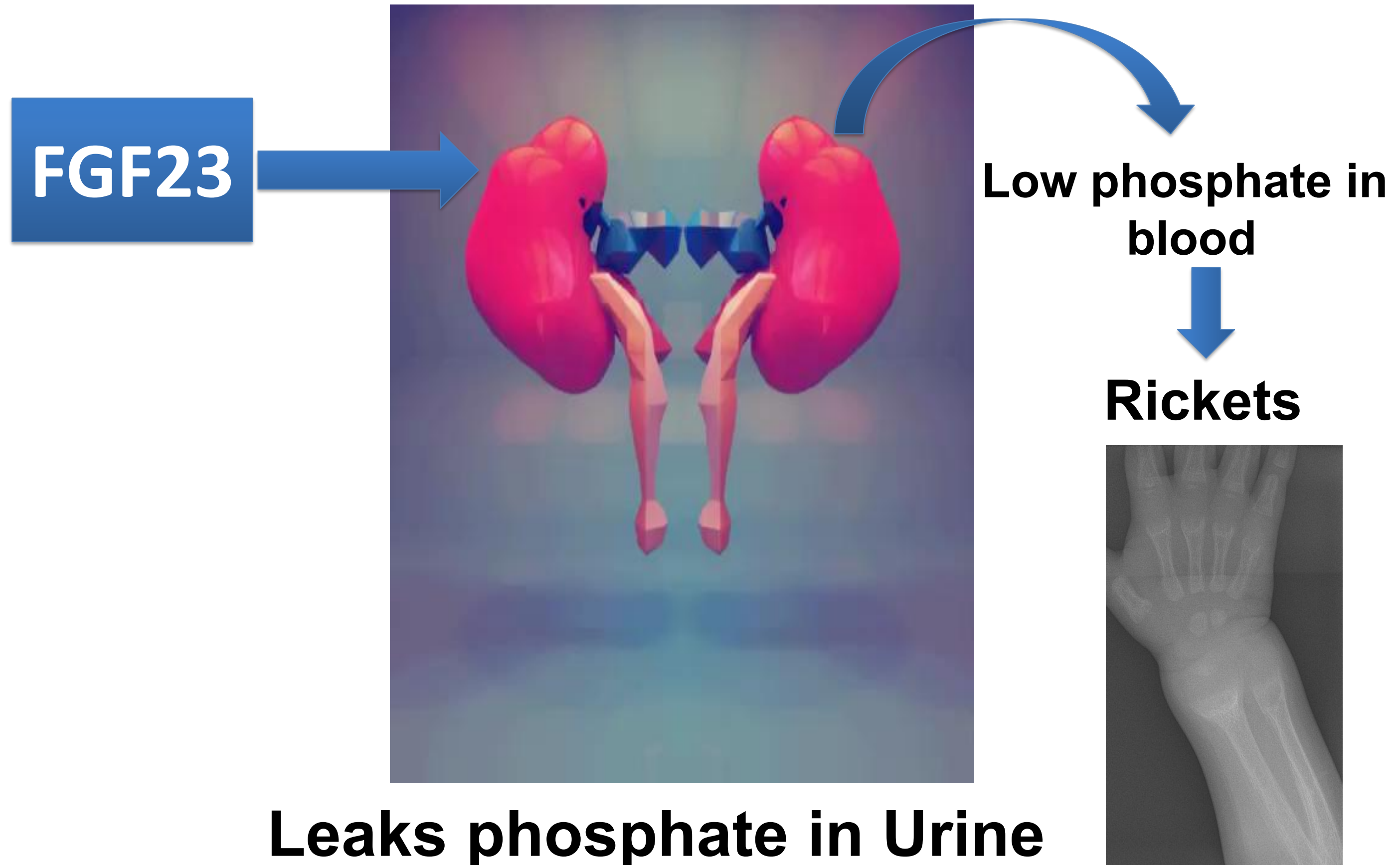
X-LINKED DOMINANT



X-LINKED DOMINANT



What does excess FGF23 in XLH do?

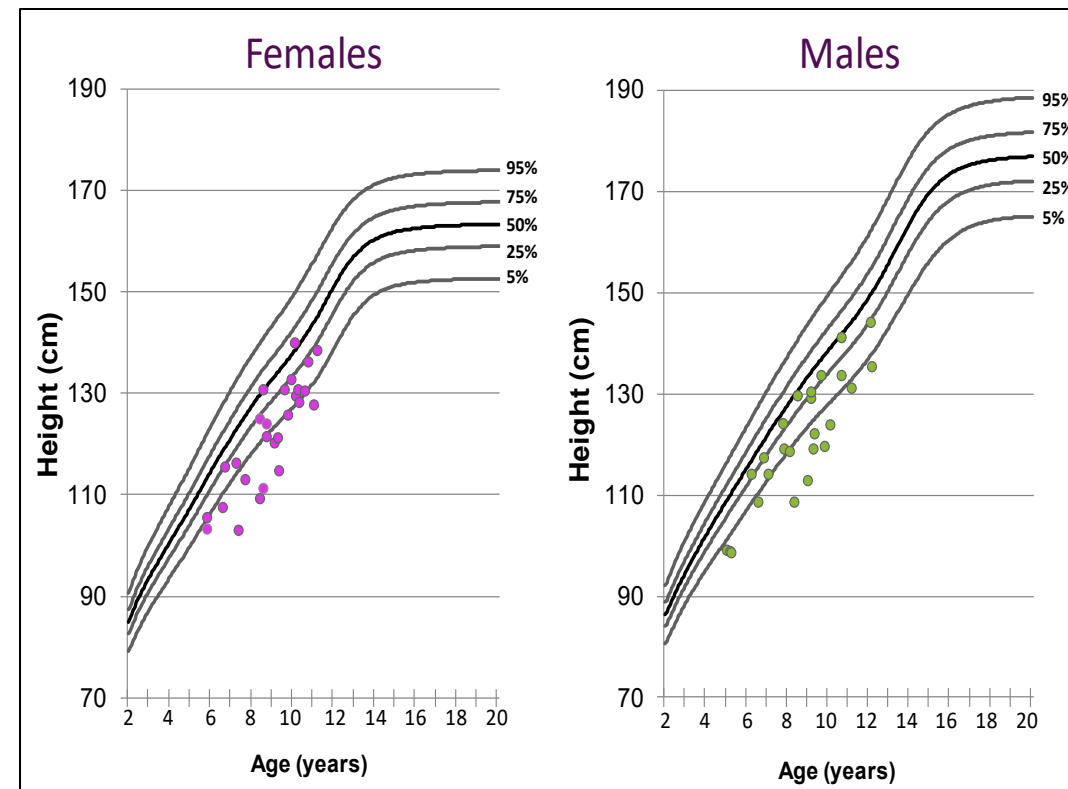


XLH in Children is Characterised by Rickets, Skeletal Deformity, Dental Abscess and Impaired Growth

Rickets



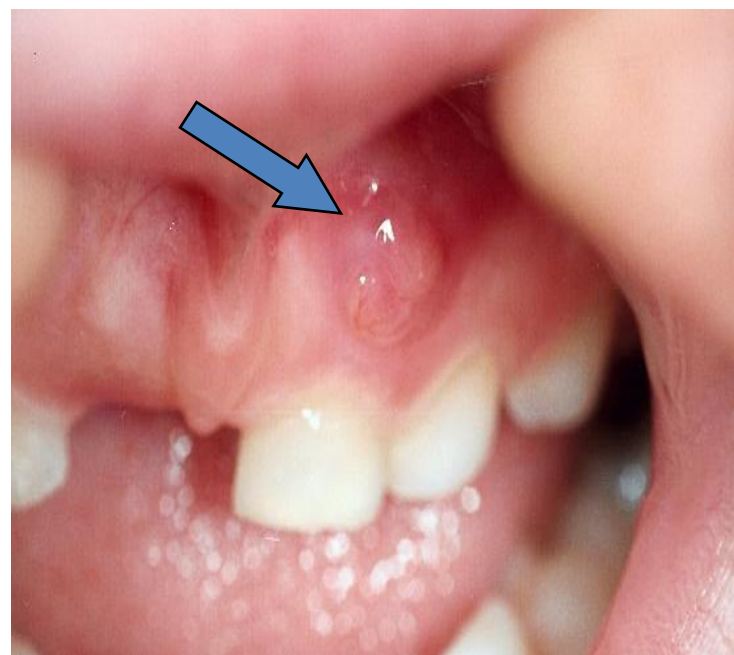
Impairment of Growth



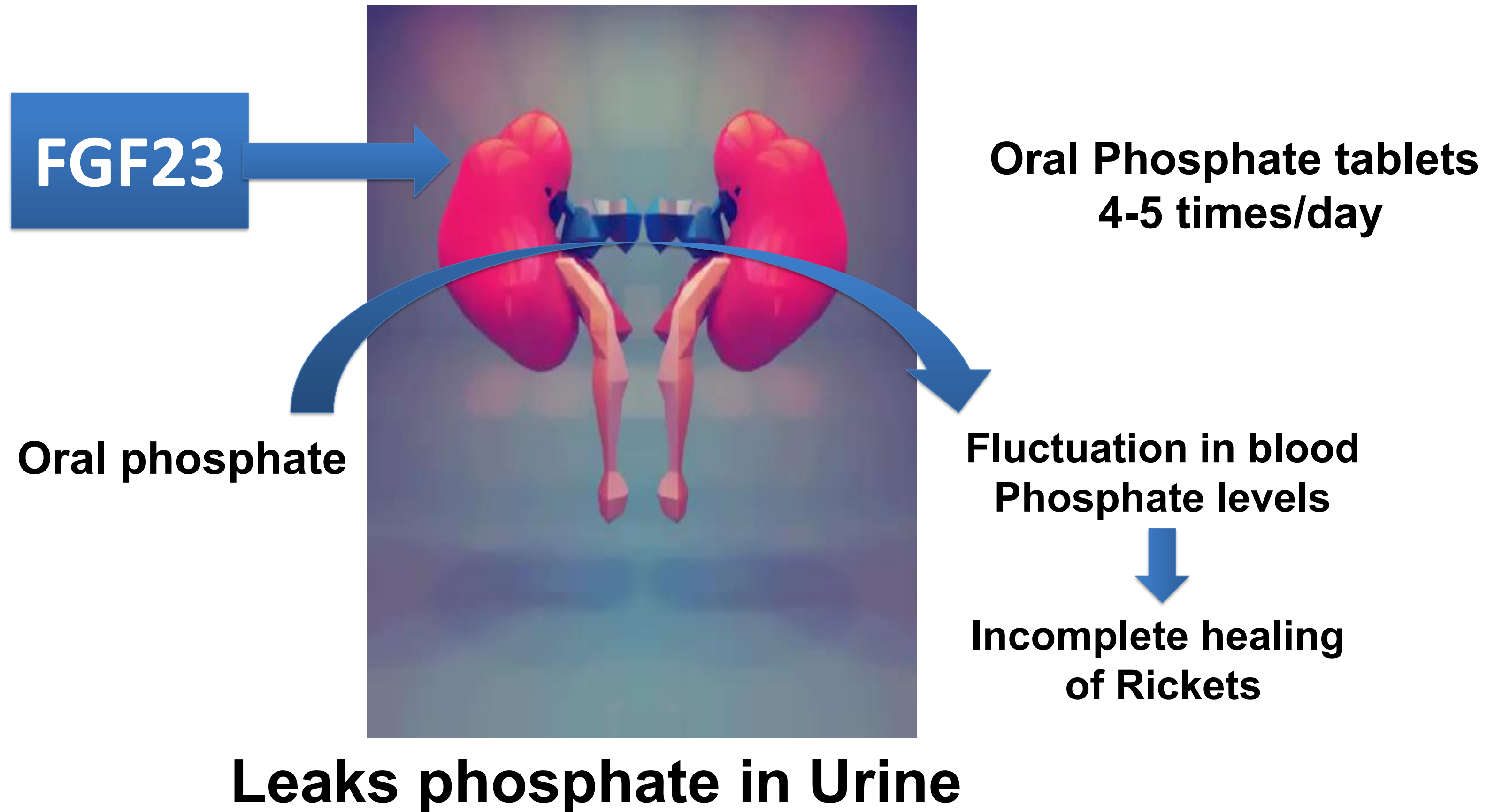
Bowing of the Leg



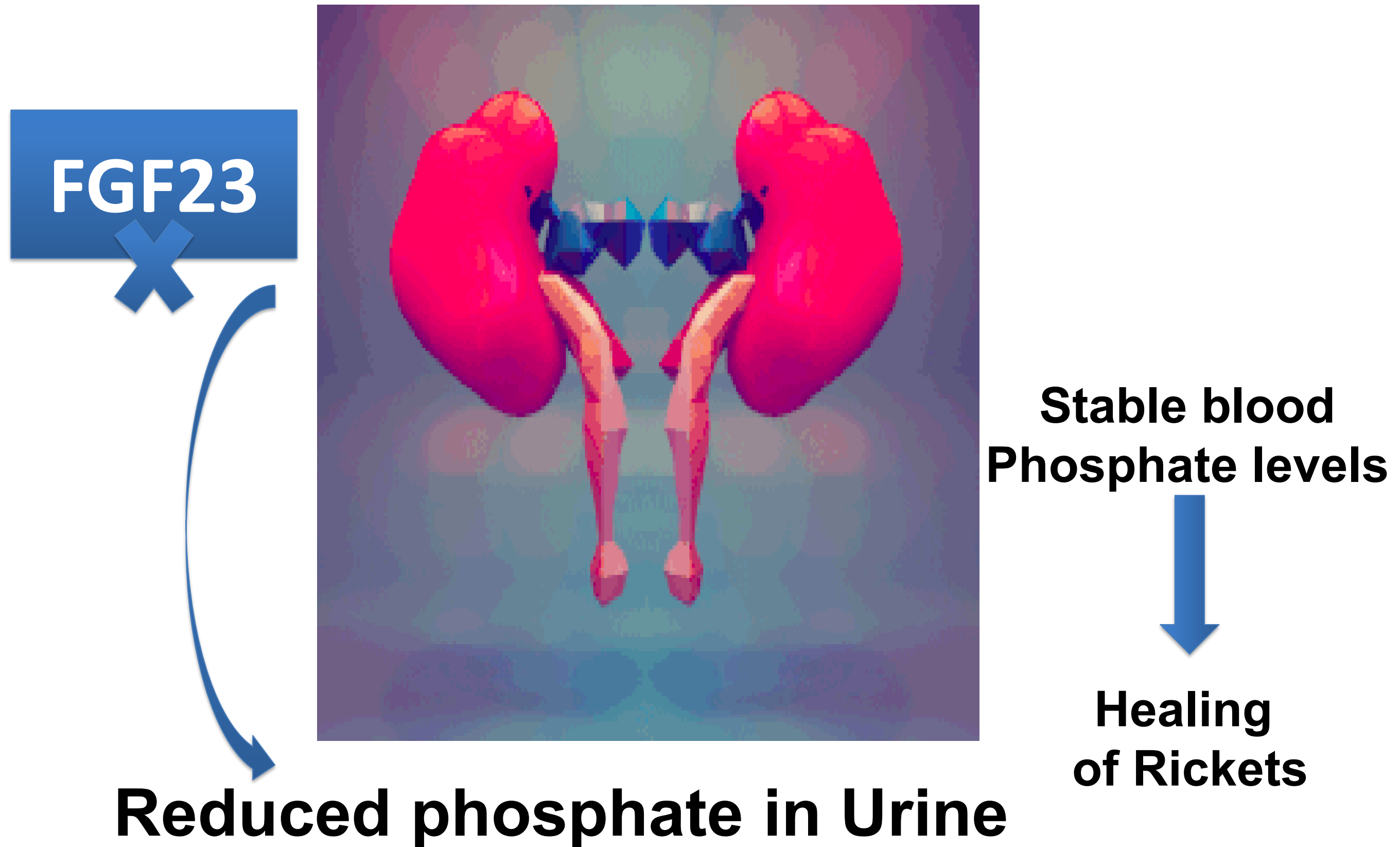
Dental Abscess



Treatment with oral phosphate

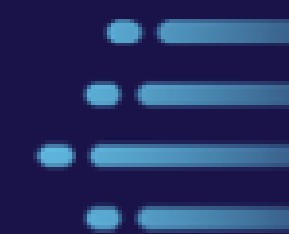


Ideal treatment would be to reduce effect of FGF23

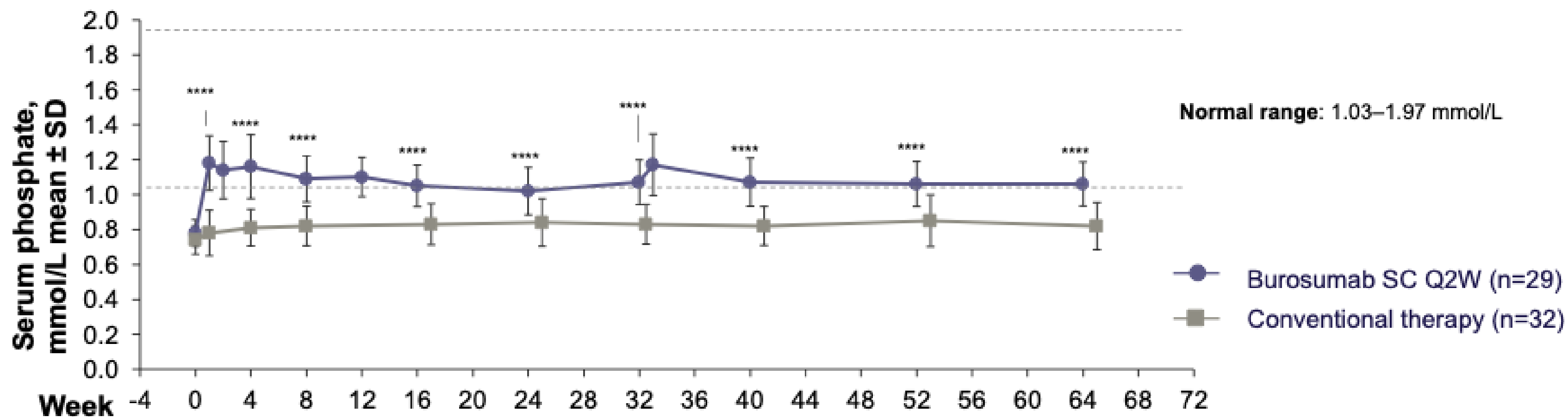


RESEARCH STUDY

Burosumab resulted in a sustained increase in fasting serum phosphate, while conventional therapy showed little change



Mean change in serum phosphate



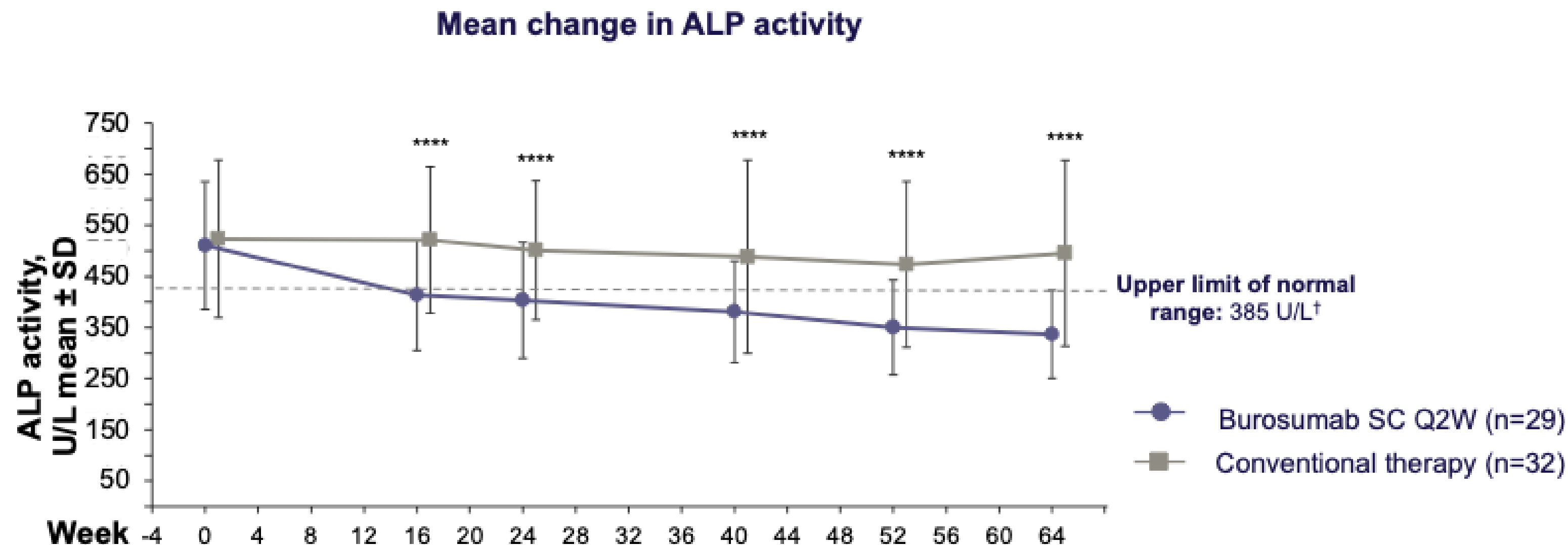
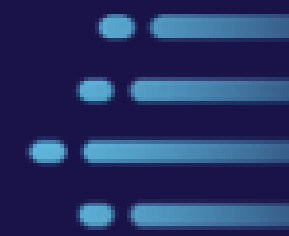
****p<0.0001 based on comparison between treatment groups in the LS mean change from baseline using a GEE model.

Some post-baseline values are slightly offset from the actual treatment week to avoid overlapping error bars.

GEE, generalised estimation equation; LS, least squares; Q2W, every 2 weeks; SC, subcutaneous; SD, standard deviation.

Imel EA, et al. *Lancet*. 2019;393:2416–27.

Burosumab resulted in a decrease in alkaline phosphatase compared to conventional therapy



****p<0.0001 based on comparison between treatment groups in the LS mean change from baseline using the GEE model. [†]Normal range varies depending on sex and age within males and females ages 1–15 years; the upper limit ranged from 297 to 385 U/L.

Some post-baseline values are slightly offset from the actual treatment week to avoid overlapping error bars.

ALP, alkaline phosphatase; GEE, generalised estimation equation; LS, least squares; Q2W, every 2 weeks; SC, subcutaneous; SD, standard deviation.

Imel EA, et al. *Lancet*. 2019;393:2416–27.

Radiological changes after Burosumab treatment



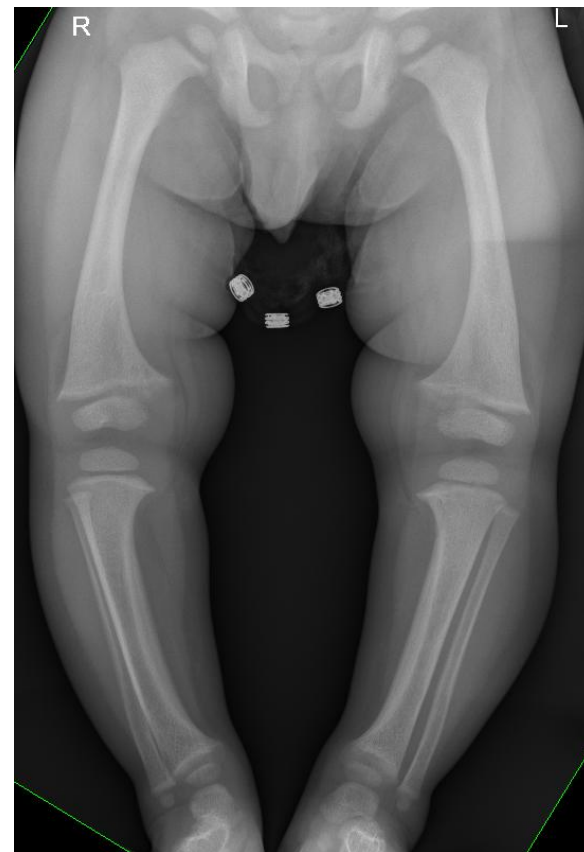
Images courtesy of Dr Padidela.

Age: 1 year
10 months.
**Pre-
treatment**



Age: 3 years
4 months.
**18 months
with
Burosumab**

Research Study: Standard treatment followed by switchover to Burosumab



Age: 1.8 years
Started 64 weeks of conventional
treatment in a Phase III clinical trial



Age: 3 years
Stopped conventional treatment
Started burosumab treatment

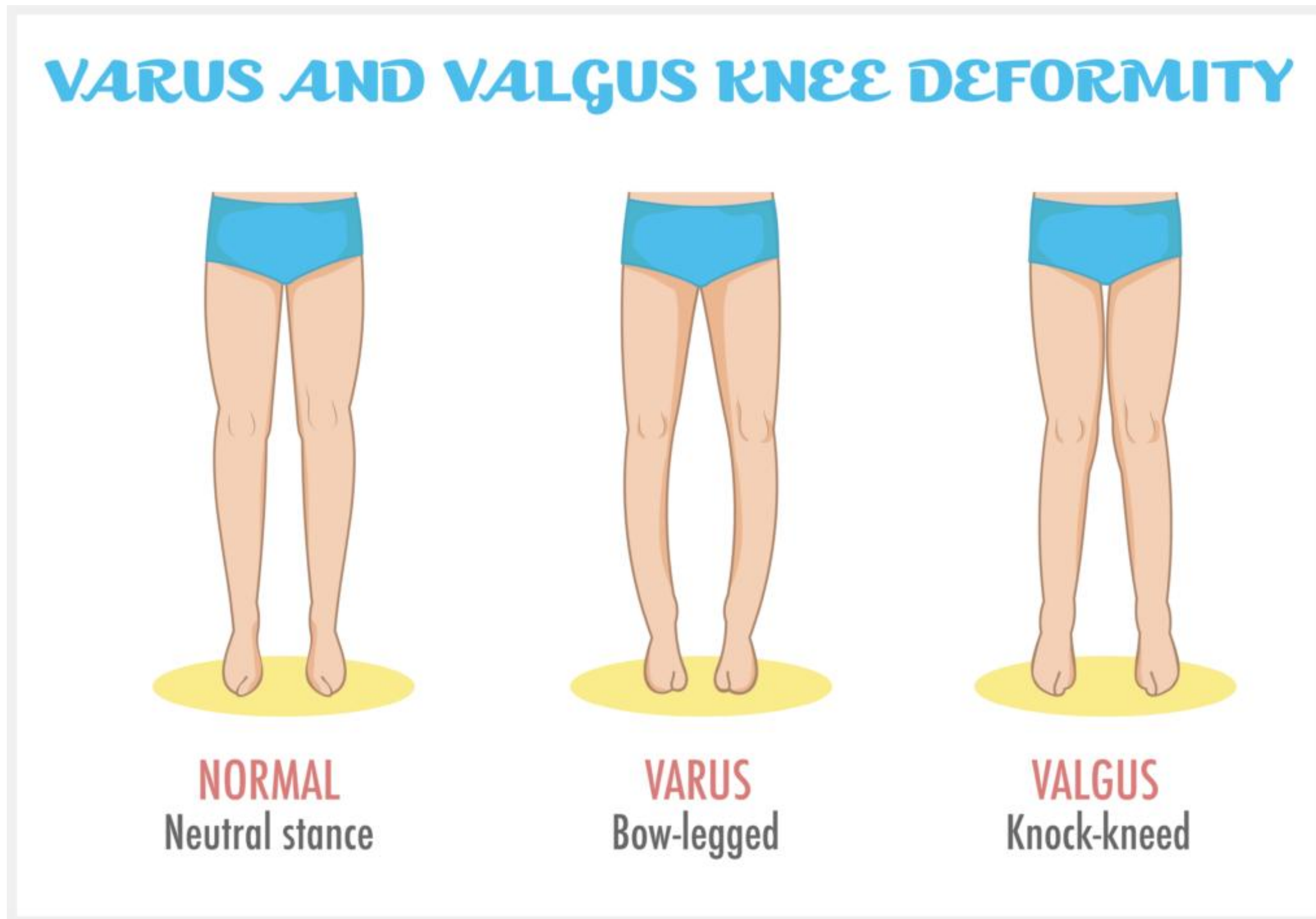


Age: 6.8 years
After 3.8 years of
burosumab treatment

Current standard of care of XLH

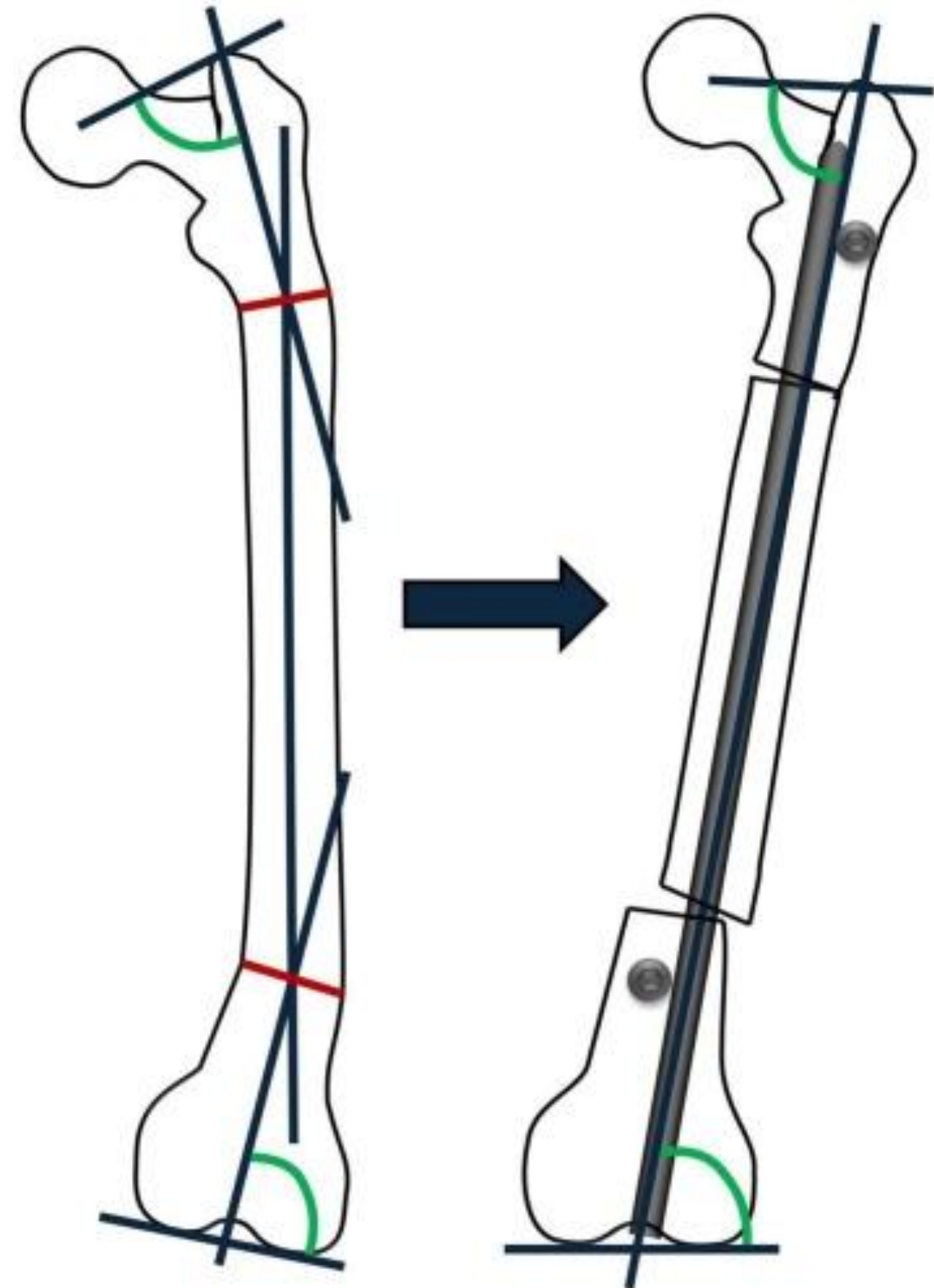
- Children younger than 1-year of age: Oral phosphate and alfacalcidol
- Children older than 1-year if age: Burosumab injections once every 2 weeks
- Adolescent after they stop growing: Oral phosphate and alfacalcidol
- Adults: Burosumab injections once every 4 weeks

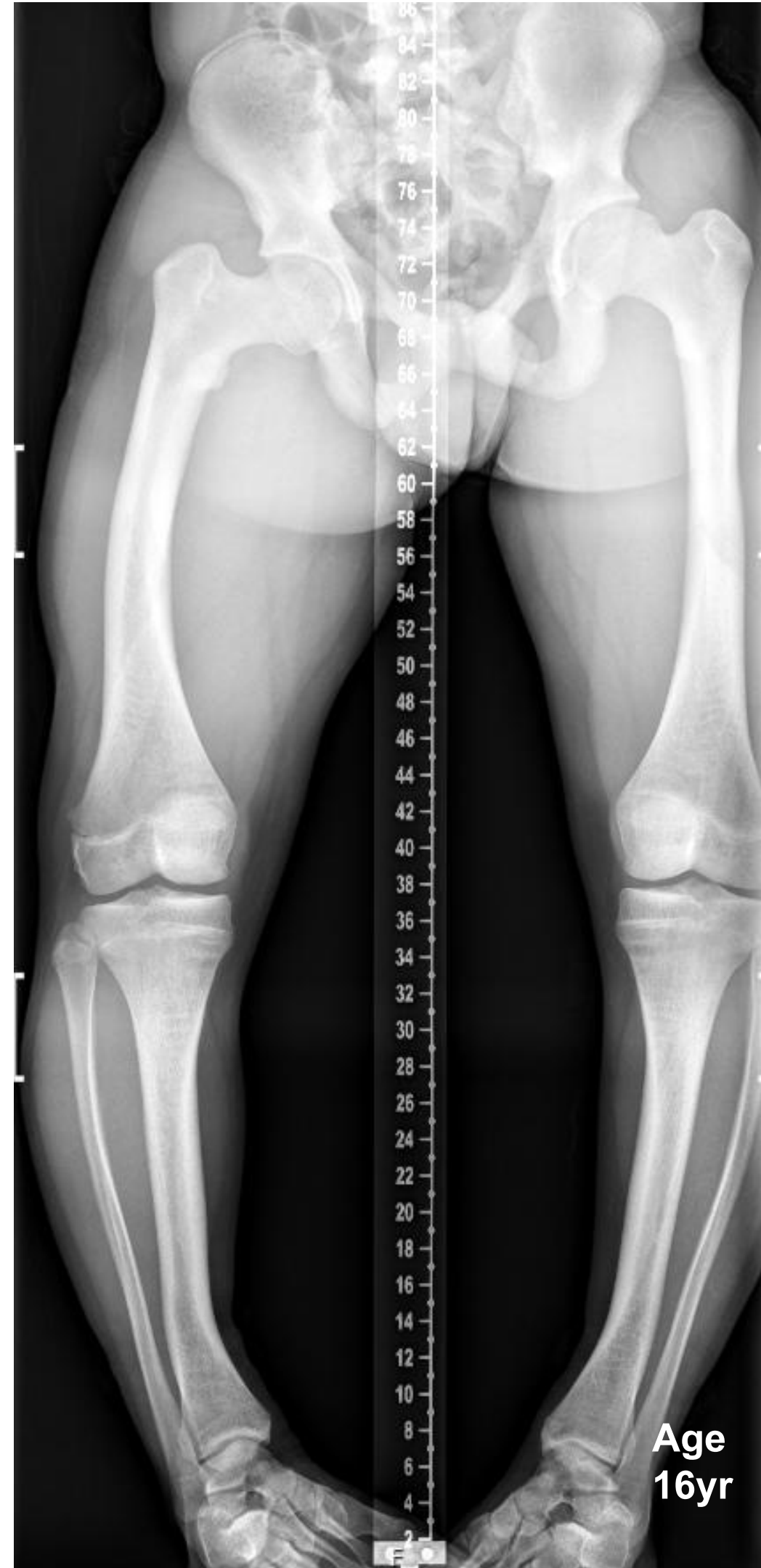
Bone Deformities in XLH



LIMB DEFORMITY CORRECTION IN XLH

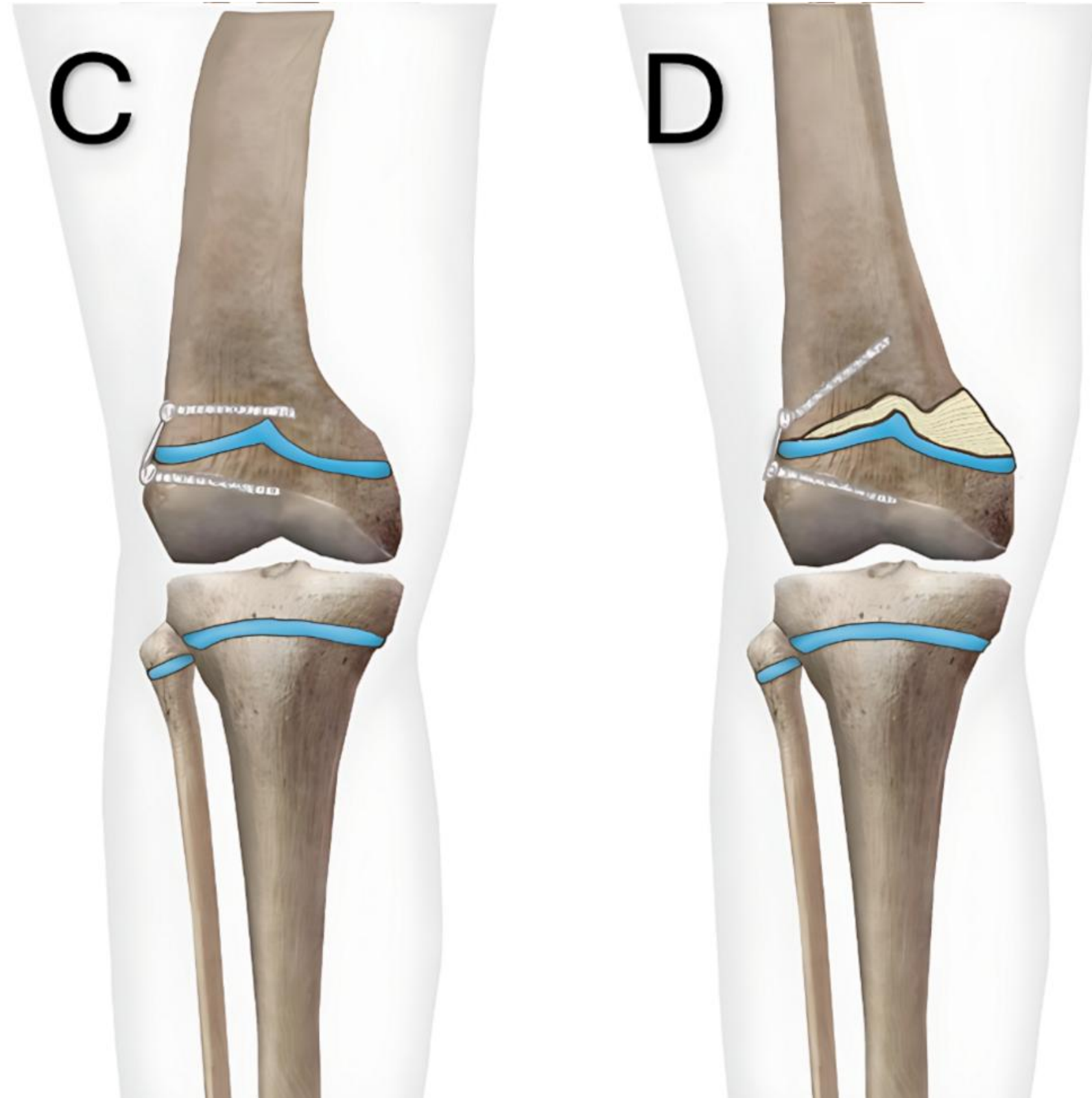
Correction of Limb Deformities: Osteotomies



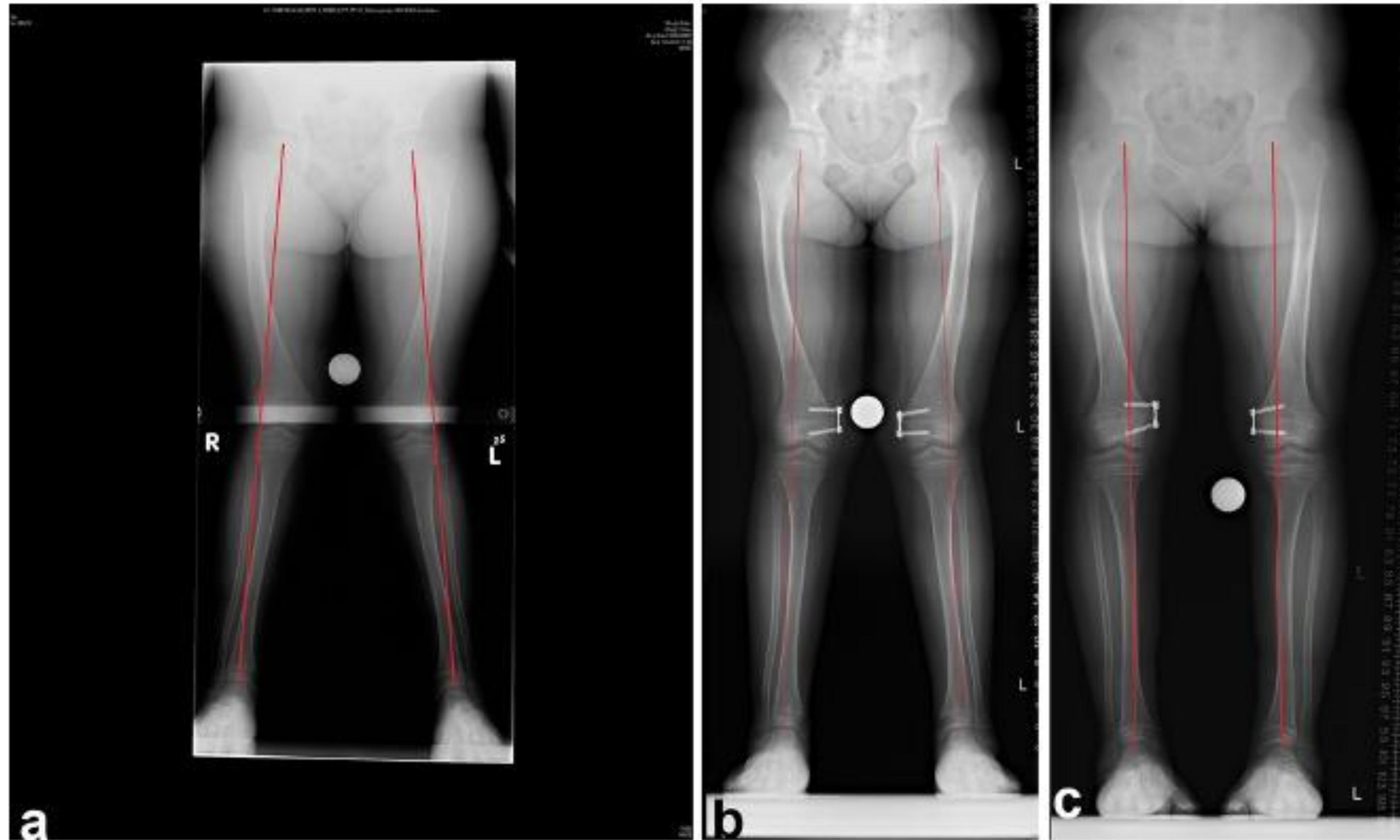




Guided growth procedure



Surgery: Knocked Knee (Genu Valgum)



Surgery: Bowed Legs(Genu Varum)



Adult XLH: bones and walking

Fractures and pseudofractures

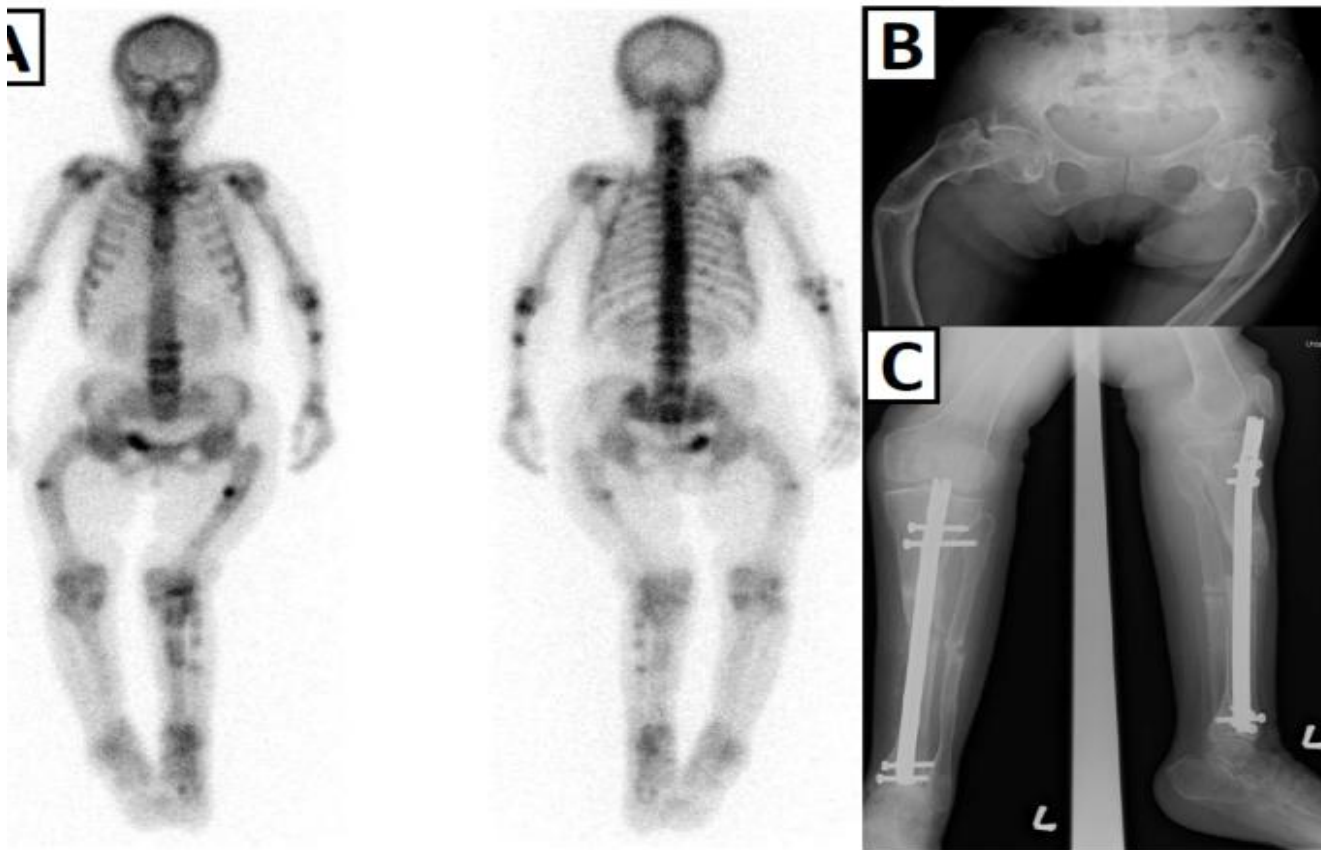
Soft, poorly mineralised bone (osteomalacia) can develop painful painful cracks, often in the legs.

Bowed legs and altered walking

Persistent leg curvature, muscle weakness and pain can make walking walking and stairs harder.

Early joint wear

Hip and knee damage can cause stiffness, reduced range of movement movement and chronic pain.



Adult XLH: joints, tendons and spine



Enthesopathy

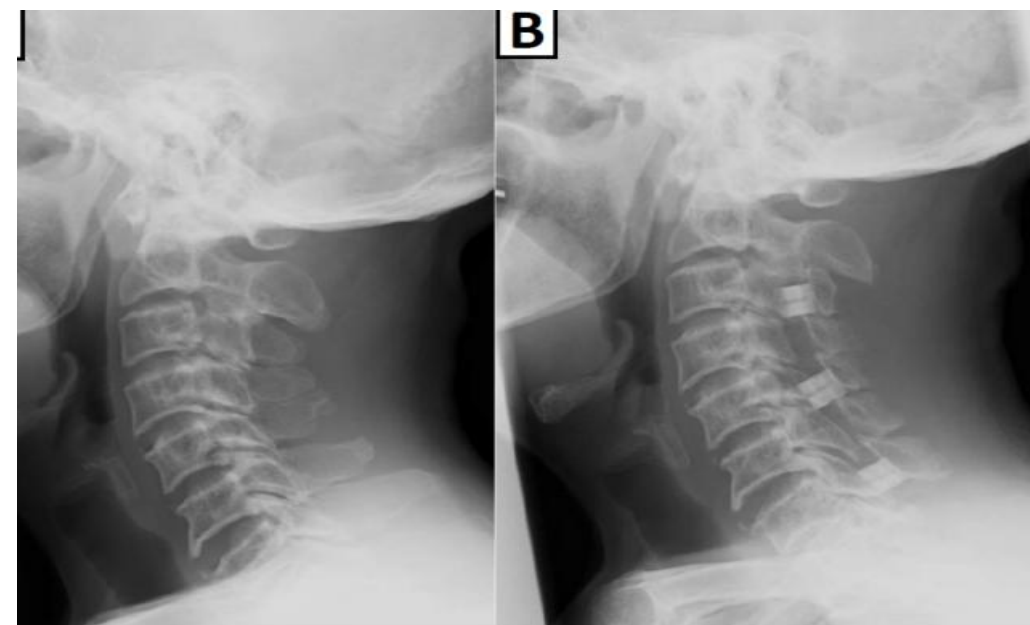
Extra bone can form where tendons and ligaments attach, contributing to pain and restricted movement.

Spinal ligament ossification

Ligaments can harden into bone, narrowing space for nerves and sometimes causing numbness or walking problems.

Osteoarthritis and stiffness

Early degenerative changes may affect the spine, hips and knees.



Adult XLH: beyond bones



Dental abscesses

Teeth may develop infections even without obvious decay; repeated root-canal treatment can be needed.



Hearing difficulties

Some adults experience hearing loss; report new changes in hearing or balance.



Fatigue and pain

Persistent pain, muscle weakness and fatigue can affect work, sleep and social life.

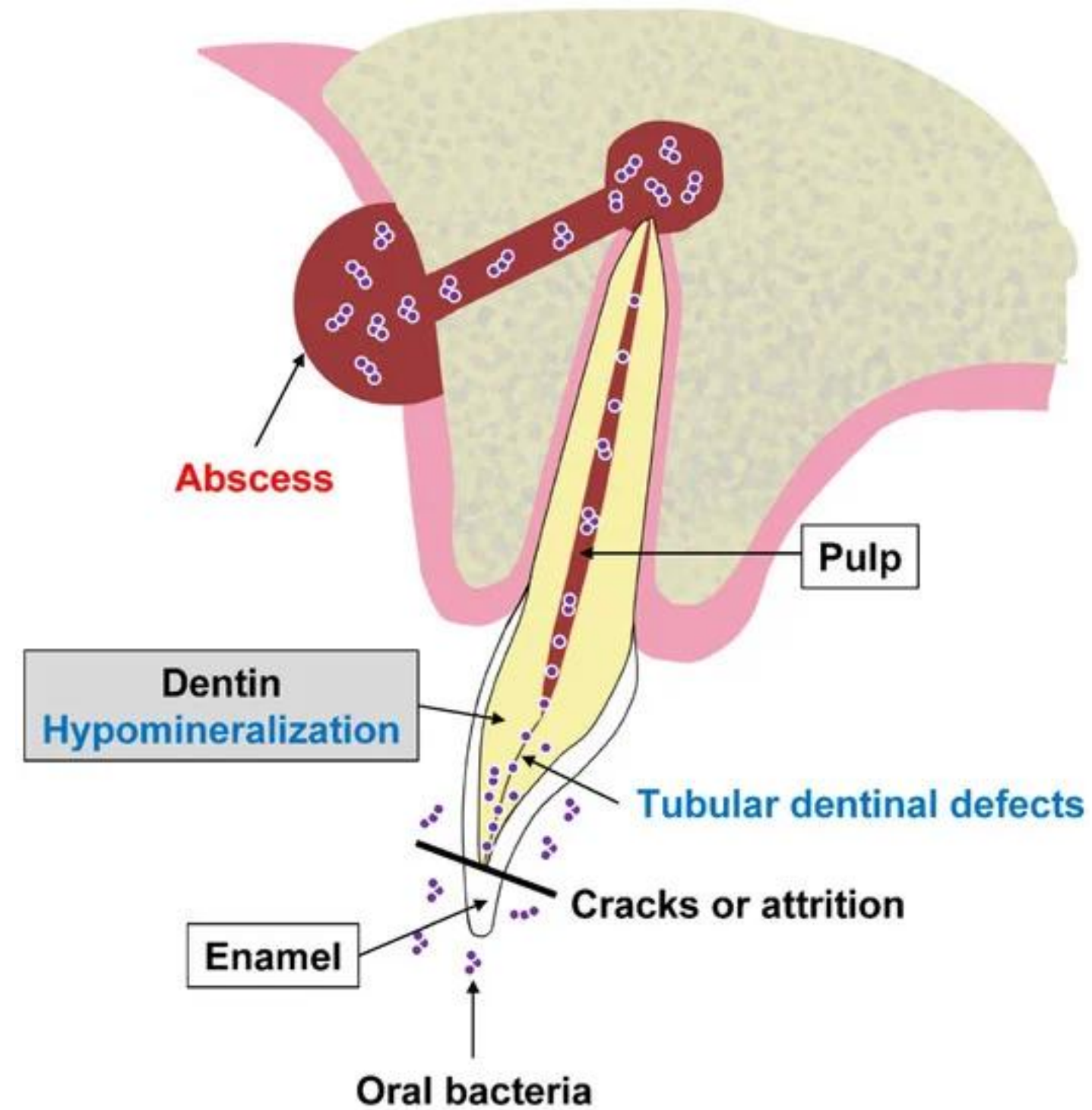


Kidney / parathyroid monitoring

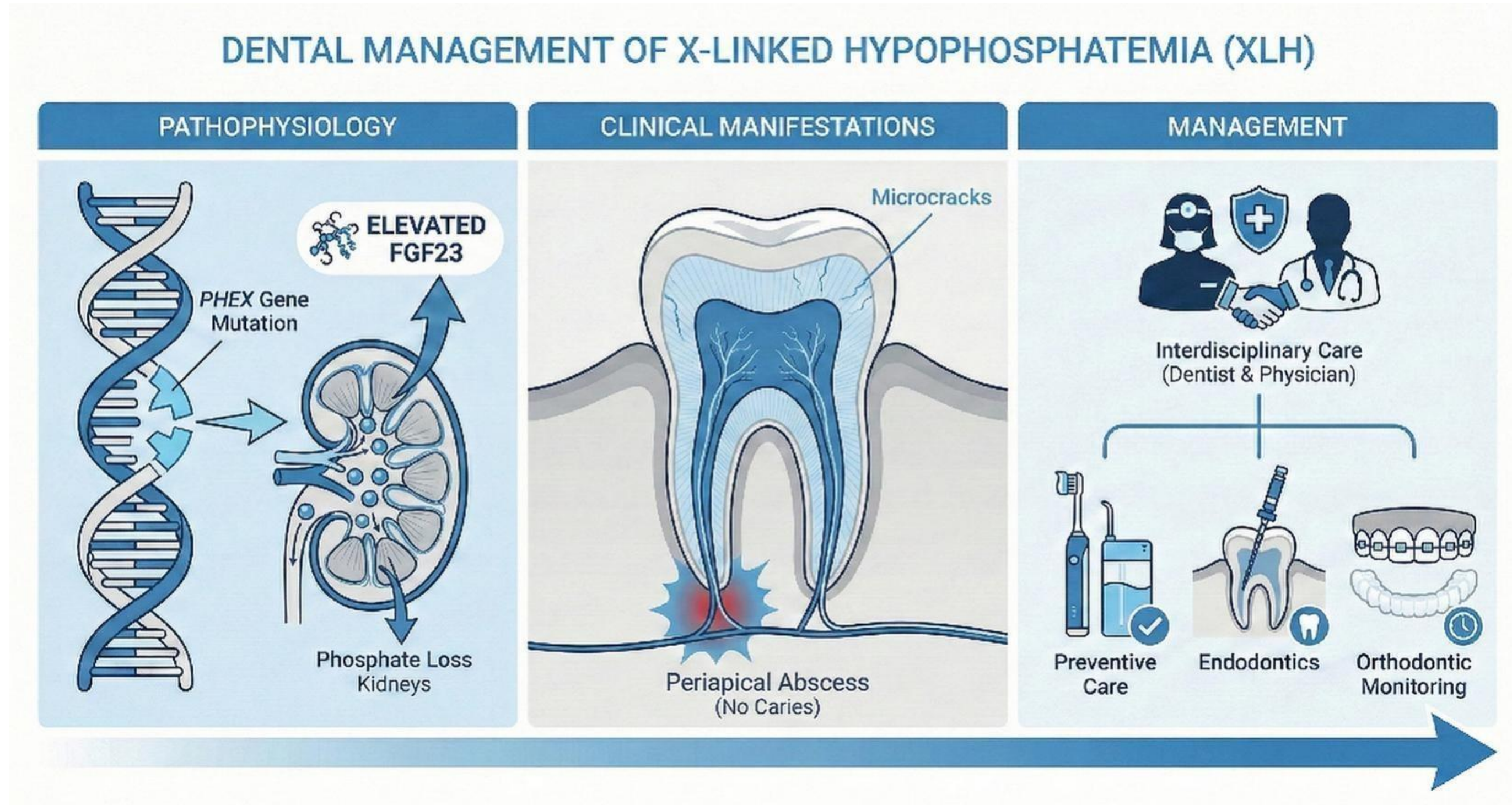
Conventional phosphate plus active vitamin D treatment can be associated with kidney calcification and high PTH.

Every person's XLH is different. Regular dental, hearing, musculoskeletal and laboratory follow-up can help.

Teeth are affected in XLH



Overview of Dental Management of XLH



Specific Dental Management

XLH: Diagnostic Guidelines and Protocol

XLH: ASSESSMENT & DIAGNOSIS

INITIAL ASSESSMENT:

- Complete medical and dental history.

CLINICAL EXAMINATION:

- Hard/Soft Tissue & Pulp Evaluation.
- Pulp Vitality Tests (Cold, Heat).
- Transillumination; Periodontal Probing.
- Assess Defects & Risk of Pulp Necrosis.

RADIOGRAPHIC EXAMINATION:

- Panoramic, Periapical, Bite-wing & CBCT.
- Identify caries, pulp/dentin defects, root anomalies, periapical lesions, bone loss.
- Annual or shorter intervals.

MONITORING OF GROWTH & FACIAL DEVELOPMENT:

- Monitor facial bone growth & dental eruption (up to 18 yrs).
- Early identification of Orthodontic & TMJ issues.
- Guides preventive treatment plan.



TREATMENT

PREVENTIVE TREATMENT



Oral hygiene instructions and dietary guidance.
Chlorhexidine mouth rinses.
Use occlusal guards.

RESTORATIVE TREATMENT



Repair enamel cracks/wear.
Treat dentinal defects; preserve pulp.
Use self-etch system.
Repair exposed dentin promptly.



ENDODONTIC TREATMENT



Pulp tests & fistula check.
Root canal: Ca(OH)_2 / bioceramic.
Antibiotics if needed.
Follow-up: radiographs & exams.



PERIODONTITIS TREATMENT



Measure periodontal pockets every visit.
Gentle root scaling & smoothing.
Chlorhexidine rinses ± antibiotics.
Avoid open-field surgeries.



SURGERY & IMPLANTS



Comprehensive pre-surgical assessment.
Physician consultation for safety.
Minimally invasive techniques.
Use PRF for healing.
Ensure hemostasis & post-op monitoring.



Thank You

Questions ?

XLHuk.org

Dr Fiona Kirkham-Wilson



CLINICAL ACADEMIC
Rheumatology

University of Oxford, NDORMS

Welcome to

RUDY

**Individuals and researchers
working together for better
rare disease care**

The RUDY study

Dr Fiona Kirkham-Wilson

XLH Community Day

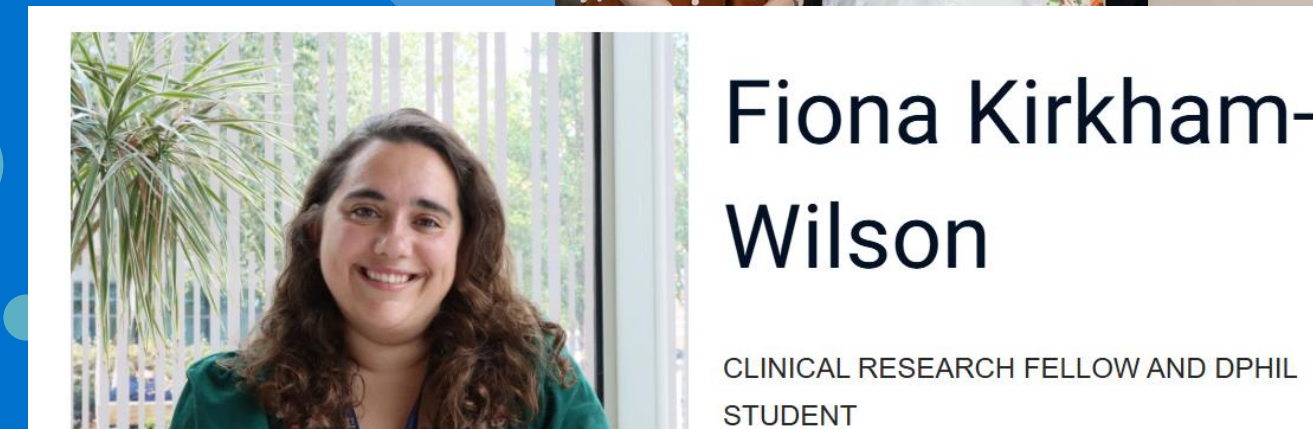
October 2026



Sign up to the
RUDY Study here

Me

- Rheumatology trainee
(doctor) – XLH Fellow
- Wife
- Mother
- Daughter
- PhD (DPhil) candidate
- Keen traveller
- Learning Japanese
- Can't reverse park or spell



Fiona Kirkham-Wilson

CLINICAL RESEARCH FELLOW AND DPHIL
STUDENT



What is the RUDY study?

- Rare and Undiagnosed Diseases Study (RUDY)
- Started 2014
- Aim: understand the experience of living with a rare bone disease



The banner features a blue background with a pattern of white and yellow dots. At the top left, it says "Transforming Clinical Care". In the center, "Welcome to" is written above the large, stylized letters "RUDY". Below "RUDY", it says "Help us improve research in rare diseases" and "Individuals and researchers working together for better rare disease care". At the bottom left, it says "Transparency in Partnership" and "Myeloma". At the bottom right, it says "Patient Driven Research". Various rare diseases are listed in circles: "Sickle Cell Disease", "XLH", "Fibrous dysplasia", "Vasculitis", "Osteogenesis Imperfecta", and "Scurvy".

NHS
National Institute for Health Research

We now have 4473 people taking part

[Find out more](#)

[↓ Scroll down for more information](#)

My role with RUDY

- Recruit participants with XLH
- Work with people with XLH to understand their priorities
- Understand how burosumab changes life for people with XLH (good and bad!)
- Who helps (cares) for people with XLH – what are their lives like? Has burosumab changed their lives?

Funded by a departmental
grant from Kyowa Kirin

What I've done

- Met lots of you!
 - XLH Connect
 - Clinic visits
 - Today
- Re-designed the RUDY platform
- Spoken about RUDY findings with other professionals
- Started work on some of the highlighted priorities
 - Women's health
 - Transition arrangements for burosumab

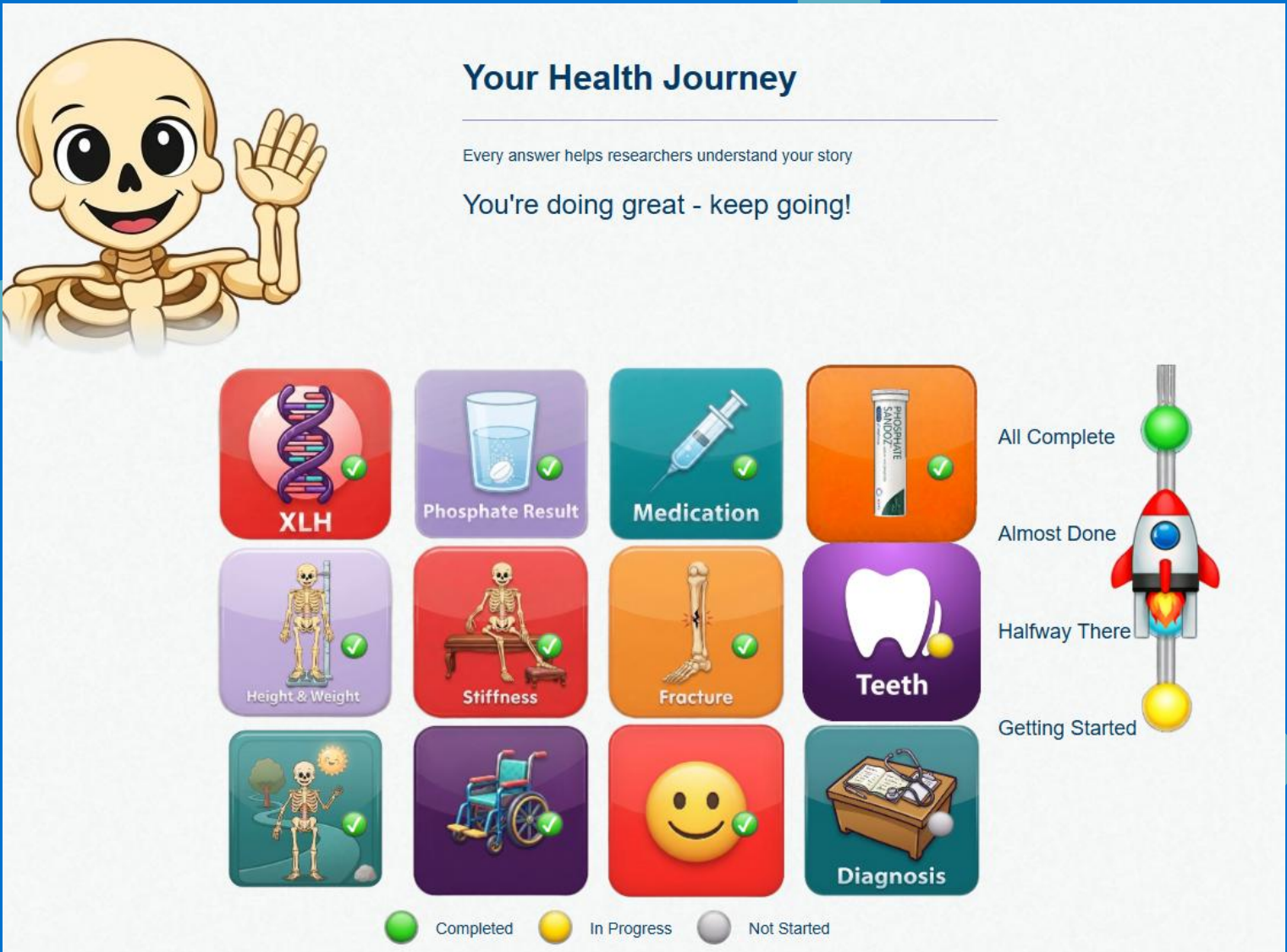
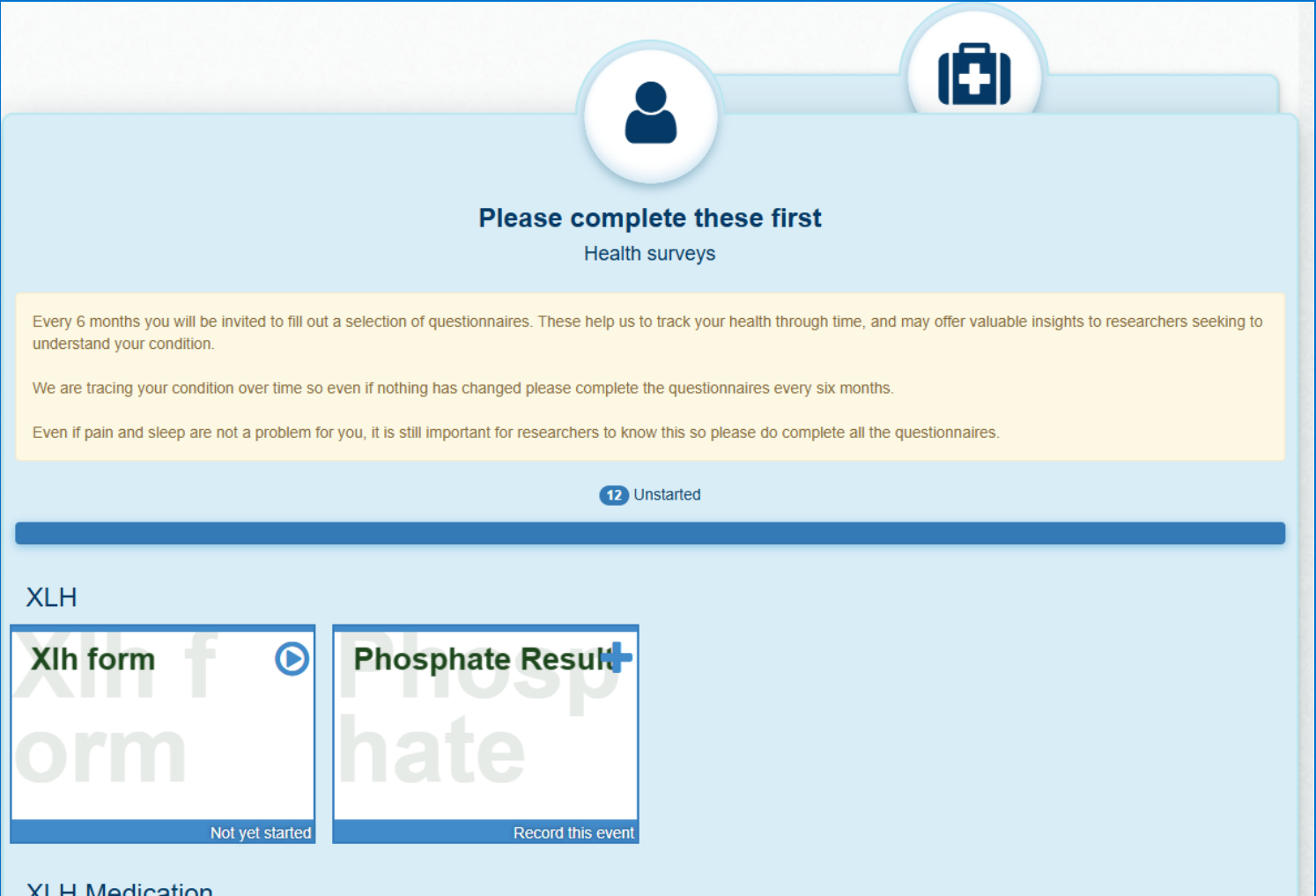
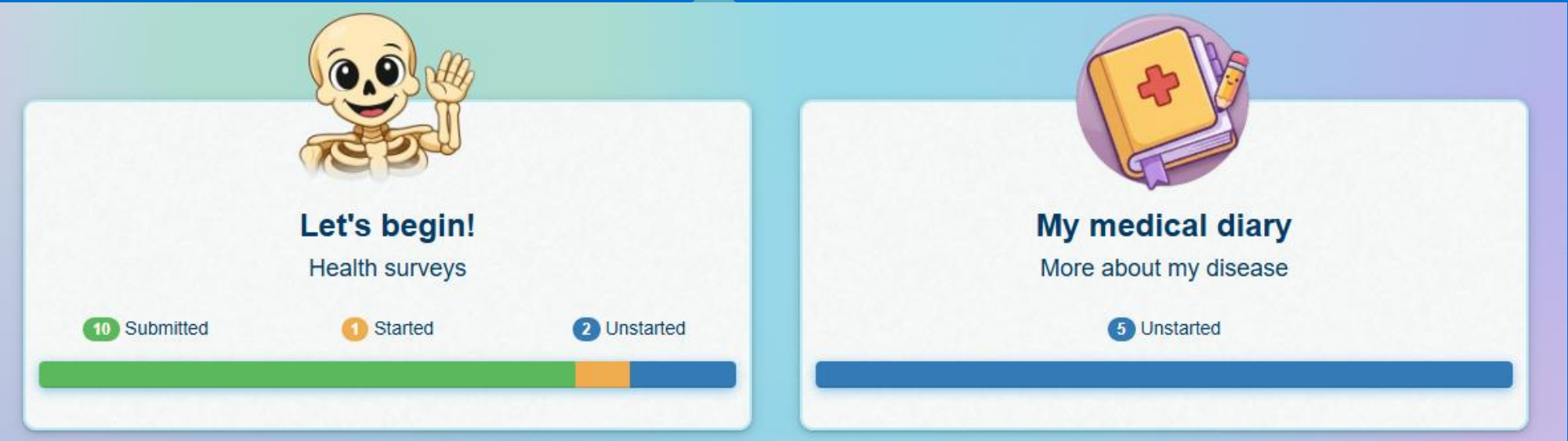


The RUDY redesign – under 16s

Before



After



The RUDY redesign – over 16s

Before

The 'Before' interface features two main sections. The first section, 'Please complete these first', is titled 'Health surveys' and shows a progress bar with '12 Unstarted'. The second section, 'My medical diary', is titled 'More about my disease' and shows a progress bar with '5 Unstarted'. At the bottom, a footer states: 'You last logged in on: 14/08/2026' and 'You have completed a total of 0 questionnaires!'.

After

The 'After' interface features two main sections. The first section, 'Please complete these first', is titled 'Health surveys' and shows a progress bar with '20 Submitted', '1 Started', and '1 Unstarted'. The second section, 'My medical diary', is titled 'More about my disease' and shows a progress bar with '13 Unstarted'.

This screenshot shows a detailed view of the 'Please complete these first' section. It includes a header with a user icon and a medical diary icon. The main content area has a title 'Please complete these first' and a subtitle 'Health surveys'. Below this, there is a yellow box with text: 'Every 6 months you will be invited to fill out a selection of questionnaires. These help us to track your health through time, and may offer valuable insights to researchers seeking to understand your condition. We are tracing your condition over time so even if nothing has changed please complete the questionnaires every six months. Even if pain and sleep are not a problem for you, it is still important for researchers to know this so please do complete all the questionnaires.' Below the text is a progress bar with '12 Unstarted'. At the bottom, there are two cards: 'Xlh form' with a play button icon and 'Phosphate Result' with a plus icon. The 'Xlh form' card has a status of 'Not yet started' and the 'Phosphate Result' card has a status of 'Record this event'.

This screenshot shows a detailed view of the 'My medical diary' section. It features a grid of cards for various medical tests and conditions. The cards are: 'XLH' (Started), 'Phosphate test' (Entered 1 - Completed), 'XLH Medication' (Complete), 'XLH old medication' (Complete), 'Bones and joints' (Submitted), 'Height & weight' (Complete), 'Stiffness' (Submitted), and 'Fractures' (Entered 2 - Completed). Each card has an icon representing the test or condition and a status bar at the bottom.

The RUDY redesign – why?

- Improved accessibility
- Better able to work on a phone/iPad
- Under 16s questions re-written → clearer
- Moved sections to *My medical diary* (optional)
- Less text
- Aim: an app

How we're doing: who contributes to RUDY?

Multiple myeloma

972

Osteogenesis
imperfecta

300

XLH

219

Sickle cell anaemia

128

Granulamatosis
with polyangiitis

99

Giant cell atheritis

71

Eosinophilic
granulamatosis
with polyangiitis

48

How XLH compares to other conditions on RUDY (as a % of UK possible participants)

XLH: ~23.0%

**Osteogenesis
imperfecta:
~7.4%**

**Multiple
myeloma:
~5.5%**

eGPA: ~1.5%

GPA: ~1.1%

**Sickle cell
anaemia:
~0.9%**

**Giant cell
arteritis:
~0.5%**

What have we found recently?

- Joint stiffness is a major symptom of XLH
- There seem to be patterns of stiffness
- Different patterns of stiffness are associated with quality-of-life indicators to a differing degree
- Joint stiffness needs to be assessed in every clinical consultation
- Burosumab seems to help joint stiffness generally – but not hip stiffness – why?
- This data has been presented at an international meeting and will be discussed further at a UK and international conference next year

What now?

- If you have, or you help someone with, XLH – please join RUDY
- Complete your 6 monthly updates (November and April are RUDY months!)
- Email me: what do you want?

Have you completed your
RUDY?
Please fill it in today or
throughout November

Thank you

• fiona.kirkham-wilson@ndorms.ox.ac.uk



Sign up to the
RUDY Study



Information about
joining the Study

Connecting as a Community



Connect

An online forum for sharing

2nd Wednesday of each month:

- 11th November
- 9th December
- 13th January
- 10th February
- 10th March

XLHuk.org



XLHuk.org

Group Photo





Contact Information

How to reach out



<https://www.facebook.com/groups/xlhukcharity>



<https://www.linkedin.com/company/xlh-uk>



https://www.instagram.com/xlh_uk



<https://twitter.com/UkXlh>



contact@xlhuk.org



<https://xlhuk.org>