Building the Bridge:
Transition Care for Pediatric Bone Disorders

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Disclosures

- I have received consulting fees from Kyowa Kiran and Alexion
- My institution receives research funding from Ultragenyx, Calcilityx and Recordati for research projects in which I am the site principal investigator

Educational Need

- Training in skeletal health disorders vastly differs between pediatrics and adult specialties
  - Rare skeletal disorders diagnosed in childhood and historically managed by pediatric teams may have unique manifestations that arise during adulthood
- Adolescence and young adulthood is a time of transition of needs outside of medicine that can impact a young person's ability to get care
  - Developing coordinated efforts to effectively transition care for persons with skeletal fragility disorders is a need of this community

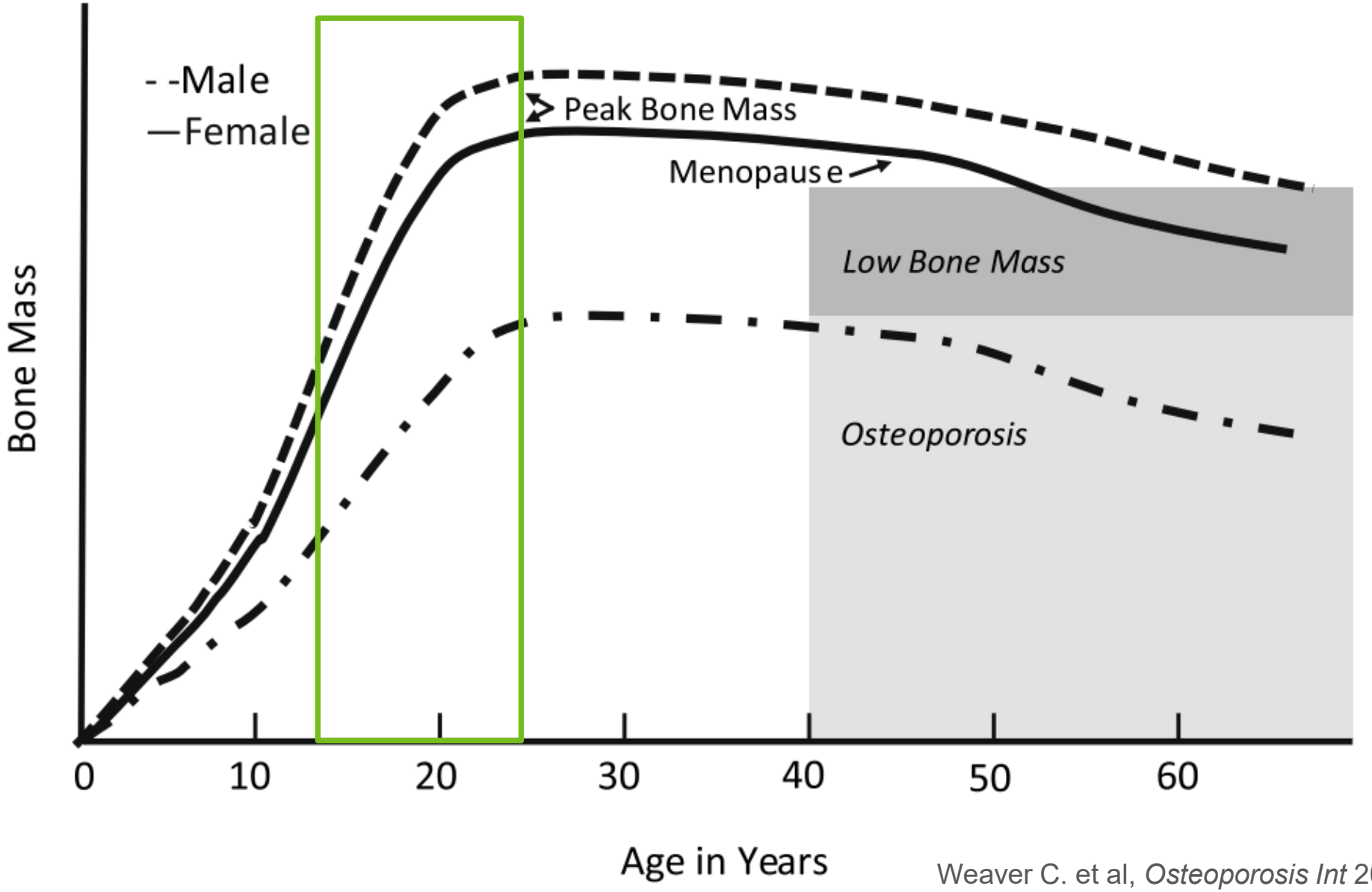
Objectives

- Examine evolving clinical needs of individuals with pediatric-onset bone disorders across adulthood highlighting disease-specific challenges that emerge during adult
- Assess gaps in current transition models and their impact on long-term outcomes for patients with pediatric-onset bone disease.
- Discuss proposed strategies to build sustainable lifespan bone health programs.

Expected Outcomes

- Stimulate discussion on how to effectively implement a transition strategy that supports the whole person and not just the medical patient
  - Identification of outcomes relevant to patients and providers that can be reliably measured to evaluate these processes

Peak Bone Mass



Weaver C. et al, *Osteoporosis Int* 2016

Who Might Need Transition Planning?

Primary Bone Disorders

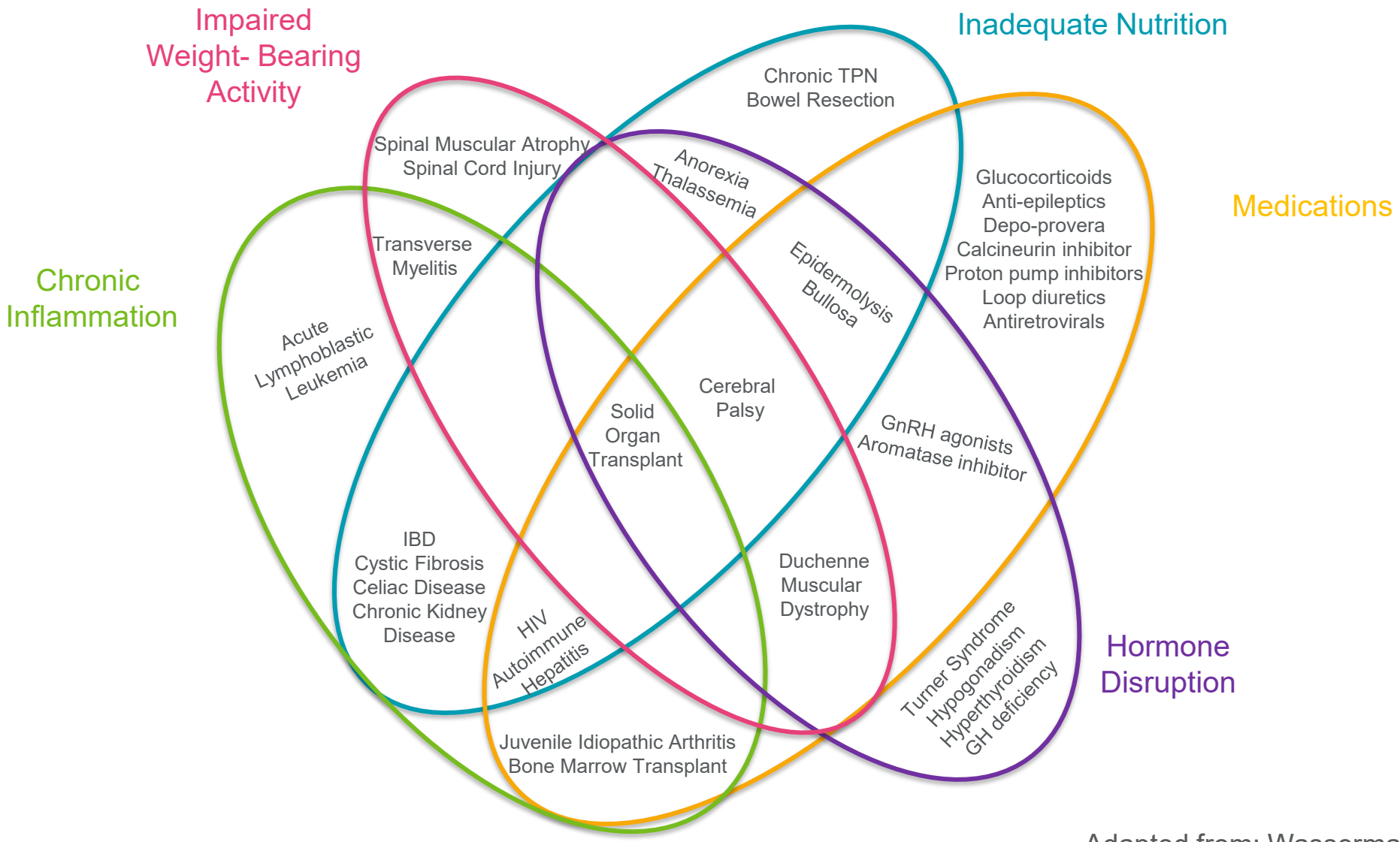
- Osteogenesis imperfecta
- X-linked hypophosphatemia
- Hypophosphatasia

Secondary Bone Disorders

- Glucocorticoid Induced Osteoporosis
- Disuse Osteoporosis

Nosology of genetic skeletal disorders 2023 revision = 771 conditions!

Secondary Osteoporosis



Adapted from: Wasserman H et al *Bone*. 2017

Changing Landscape of Pediatric Bone Health

- 25-30 years ago:
  - Many rare skeletal disorders viewed primarily as pediatric diseases
  - Limited treatment options
  - Less survival into adulthood
- Today
  - Improved diagnostics
  - Disease-specific therapies
  - Better orthopedic management
  - Improved life expectancy



1983 — Identified as causative gene for Hypophosphatasia

ALPL

1988 — **COL1/COL1A2**

Recognized as a major cause of Osteogenesis Imperfecta

1995 — **PHEX**

Identified as gene associated with X-linked Hypophosphatemia

1998 — Glorieux and colleagues published landmark NJEM study using pamidronate to treat patients with severe OI

Bisphosphonates

2000 — **FGF-23**

Identified as key phosphotonin regulator involved in XLH

2013 — Approved for treatment of osteoclast rich tumors in children with fused epiphyses

Denosumab

2015 — **Asfotase Alfa**

Approved for infantile and childhood onset hypophosphatatasia

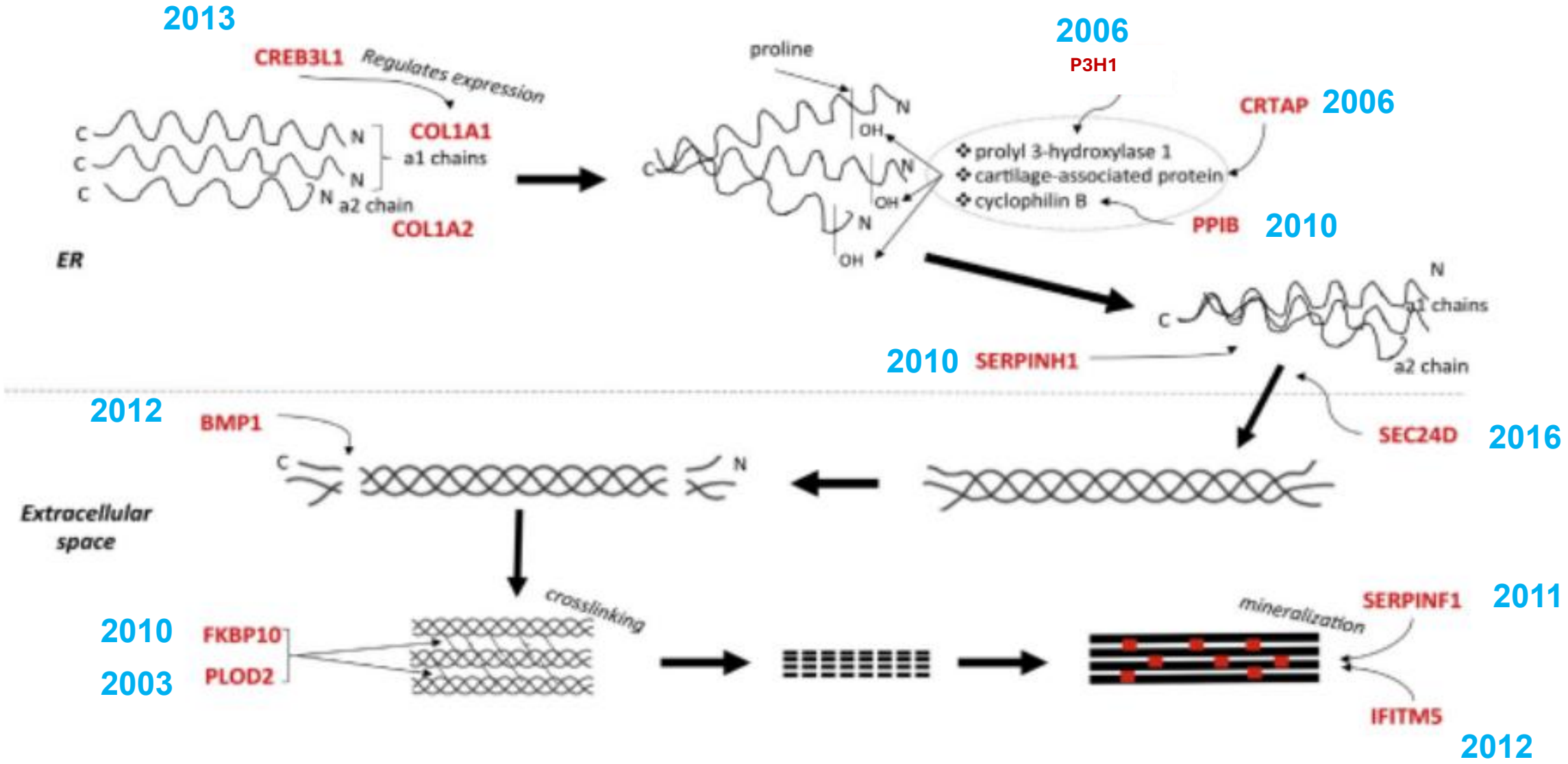
2018 — Approved for treatment of X-linked Hypophosphatemia

Burosumab

2023 — **Anabolic Agents**

Anti-sclerostin antibodies investigated for treatment of Osteogenesis Imperfecta

Major OI Gene Discoveries



Tournis S, Dede AD, *Metabolism* 2018

Transition vs. Transfer

- Transfer of Care
  - Single event
- Transition
  - Process that begins years before transfer
  - Builds self management skills
  - Establish adult care relationships

National Alliance to Advance Adolescent Health

got transition

SIX CORE ELEMENTS™ APPROACH AND TIMELINE FOR YOUTH TRANSITIONING FROM PEDIATRIC TO ADULT HEALTH CARE

1. **POLICY/GUIDE**
   Develop, discuss, and share transition and care policy/guide
   AGE 12-14
2. **TRACKING & MONITORING**
   Track progress using a flow sheet registry
   AGE 14-18
3. **READINESS**
   Assess self-care skills and offer education on identified needs
   AGE 14-18
4. **PLANNING**
   Develop HCT plan with medical summary
   AGE 14-18
5. **TRANSFER OF CARE**
   Transfer to adult-centered care and to an adult practice
   AGE 18-21
6. **TRANSITION COMPLETION**
   Confirm transfer completion and elicit consumer feedback
   AGE 18-23

Adolescent Development and Transition Readiness

Challenges

- Executive function still developing
- Emerging independence
- College or employment transitions
- Insurance changes
- Leaving home

Questions to ask

- What is my diagnosis?
- What medications do I take?
- Why do I take them?
- What happens if I stop them?
- Who do I call when problems arise?

X-Linked Hypophosphatemia

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X-Linked Hypophosphatemia

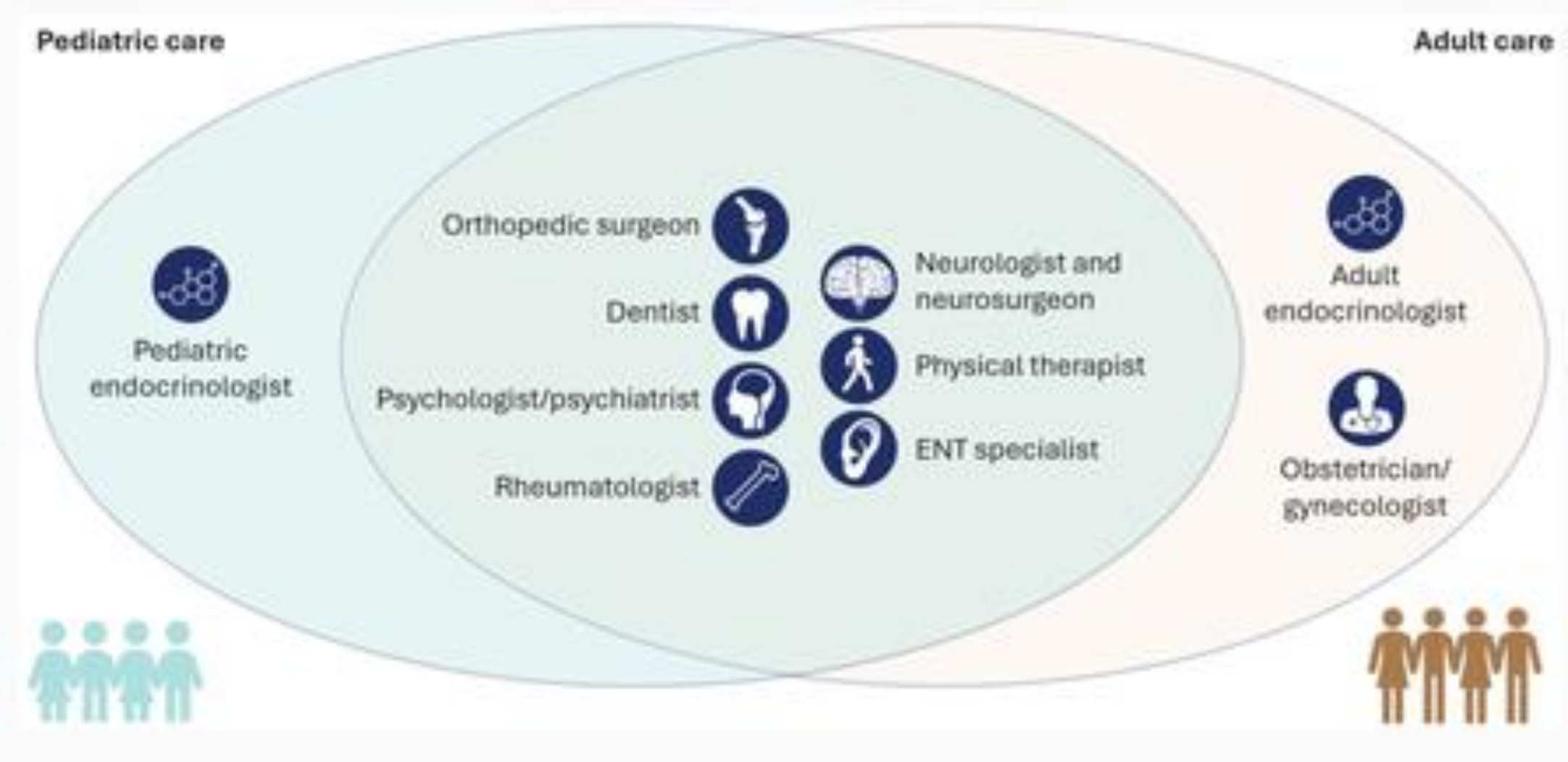
Pediatric issues

- Rickets
- Leg deformities
- Growth
- Dental Disease
- Craniosynostosis/Chiari I

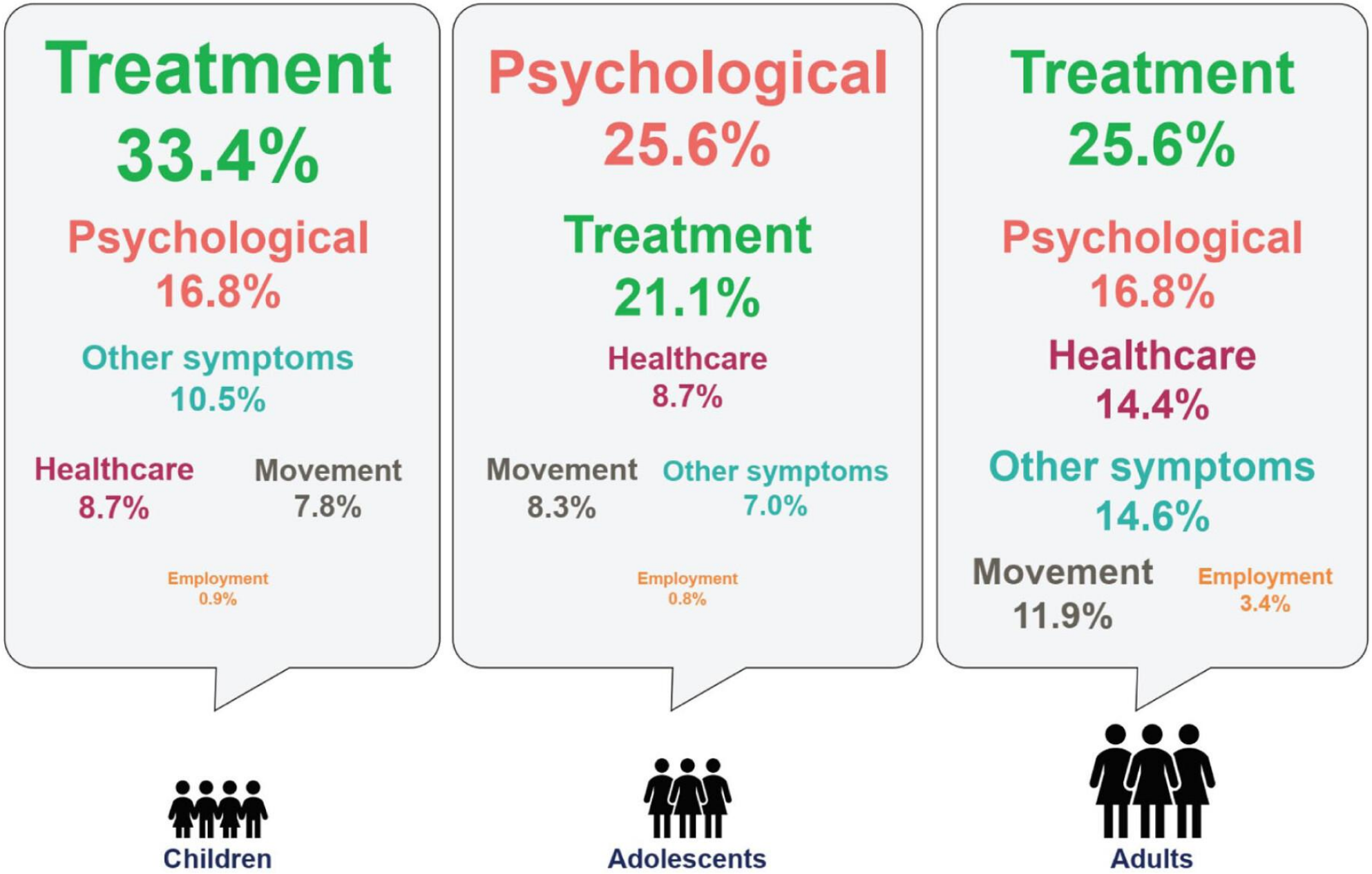
Adult Issues

- Enthesopathies
- Osteoarthritis
- Fractures/Pseudofractures
- Nephrocalcinosis
- Dental Disease
- Chronic pain
- Syringomyelia

XLH Matters



Seefried et al *Clin Med Insights Endocrinol Diabetes* 2026



Seefried et al *Clin Med Insights Endocrinol Diabetes* 2026

XLH Community Guidebook

Resources and Glossary

Understanding XLH | Managing XLH | Monitoring XLH | Key Transitions | Daily Living | Health and Wellness

Find Support

Preparing for Key Transitions

Read this section for information about preparing for key medical and life transitions.

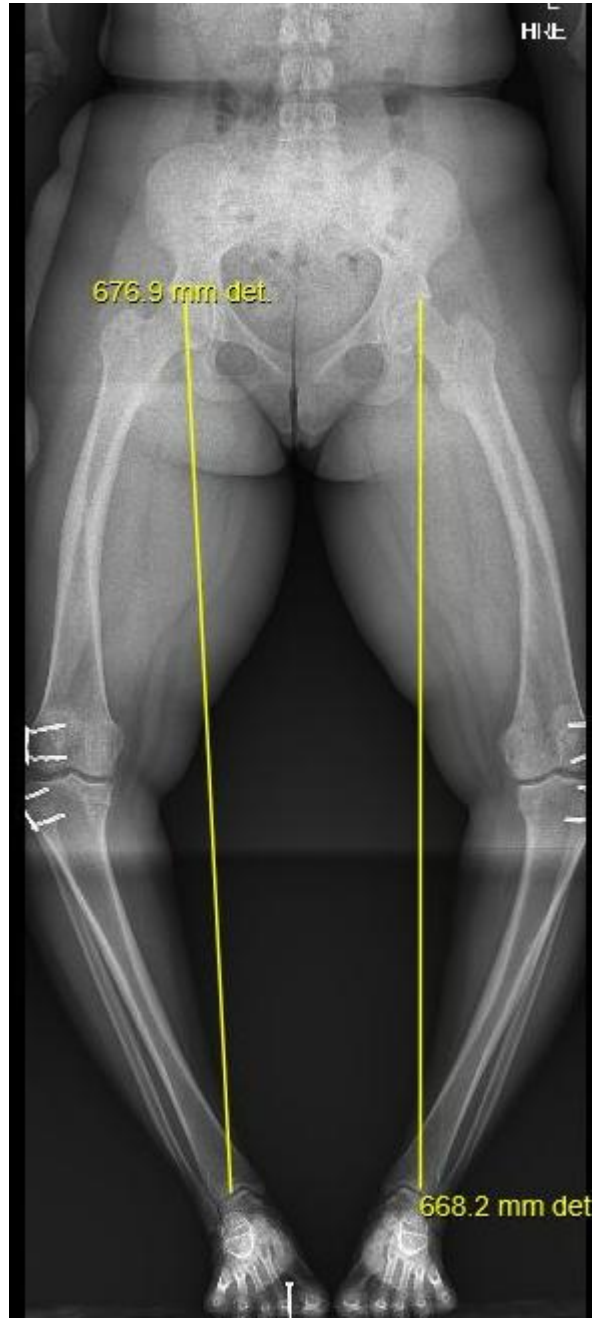


Importance of Transitions | Moving to Adult Doctors | Care Transition Process | Life Transitions

Preparing for Key Transitions - XLH Community Guidebook

Key Management Decisions Around Transition

- Medications
  - Burosumab
    - When to transition for Q2 weeks to Q4 weeks?
    - How can clinicians help young adult patients maintain routine is changing (college, work force etc)?
- Surgical Intervention
  - Guided growth vs. osteotomy
- Dental care
- Pain and stiffness



Osteogenesis Imperfecta

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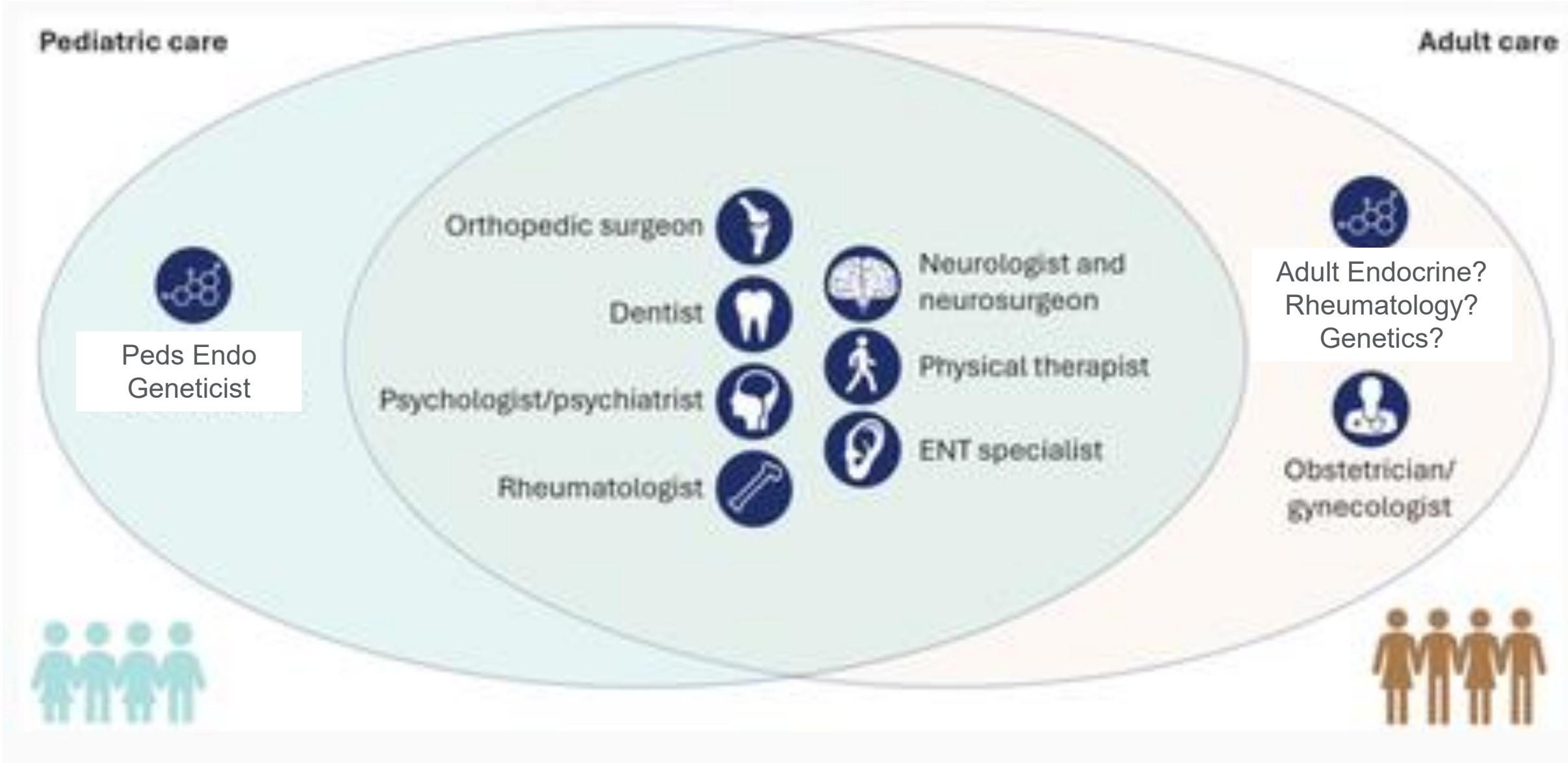
Osteogenesis Imperfecta

Pediatric Focus

- Fracture prevention
- Growth
- Mobility
- Bisphosphonate treatment – goal is to get to drug holiday

Adult Focus

- Chronic Pain
- Hearing Loss
- Osteoarthritis
- Spine Disease
- Functional decline
- Resume bisphosphonate treatment or other anabolic therapy


Pediatric care
Adult care
Peds Endo
Geneticist
Orthopedic surgeon
Dentist
Psychologist/psychiatrist
Rheumatologist
Neurologist and neurosurgeon
Physical therapist
ENT specialist
Adult Endocrine?
Rheumatology?
Genetics?
Obstetrician/ gynecologist

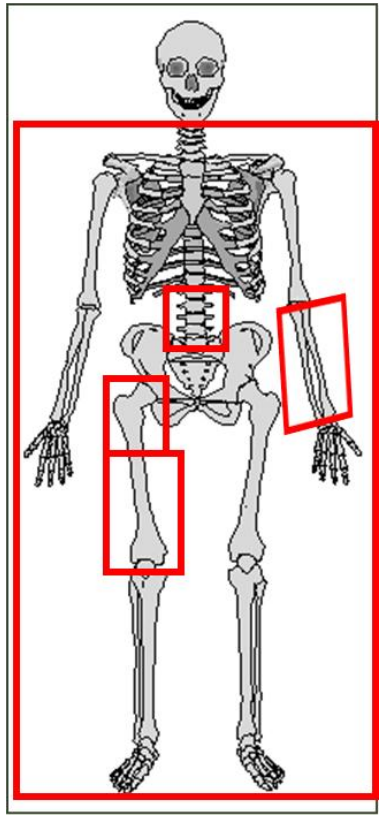
Patient Journey - OIFE

- Pediatric care: bisphosphonate infusions at regular intervals until fusion of growth plates
  - Natural history for OI Type 1 is fracture rates decrease
- Transition around age 18 – to adult medicine where there is no standard of care for adults with OI
  - Limited access to multidisciplinary teams
  - Adult patients reported feeling “overwhelmed” by having to form their own medical teams or receiving siloed care from non-experts in OI
  - Hesitancy to resume treatment if fractures are more intermittent

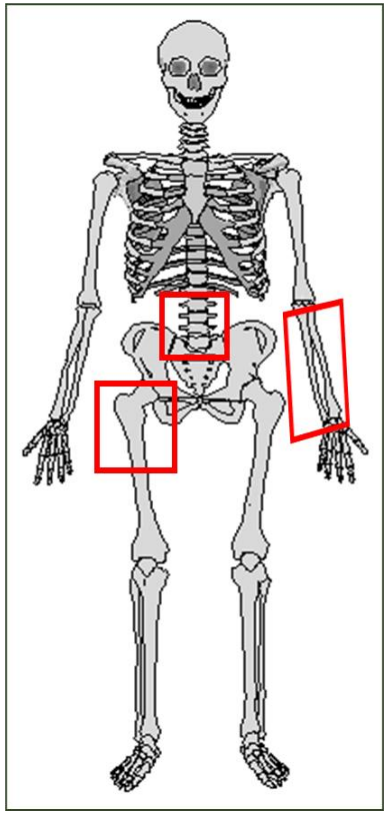
→ utilization of emergency resources

Westerheim et al. *Orphanet Journal of Rare Diseases* 2024

ISCD Recommended Body Sites (DXA)



Pediatric (2019)	Adult 2023
Lumbar Spine (PA)	Lumbar Spine (PA)
Total Body Less Head	Proximal Femur
Forearm (ambulatory)	Forearm (if unable to measure hip and/or spine, hyperparathyroidism, obesity)
Lateral Distal Femur (non-ambulatory, non-removable artifacts at other sites)	
Proximal femur (reduced weight bearing, transition to adult care)	



Key Management Decisions Around Transition

- Bisphosphonates
  - When to start a drug holiday?
  - What DXA scans to obtain to aid in both pediatric and adult monitoring of bone mineral density?
  - When to monitor bone turnover markers?
- Anabolic therapy
  - When might romosozumab or PTH analogs be beneficial for a young adult patient?

Trabecular Bone Score?

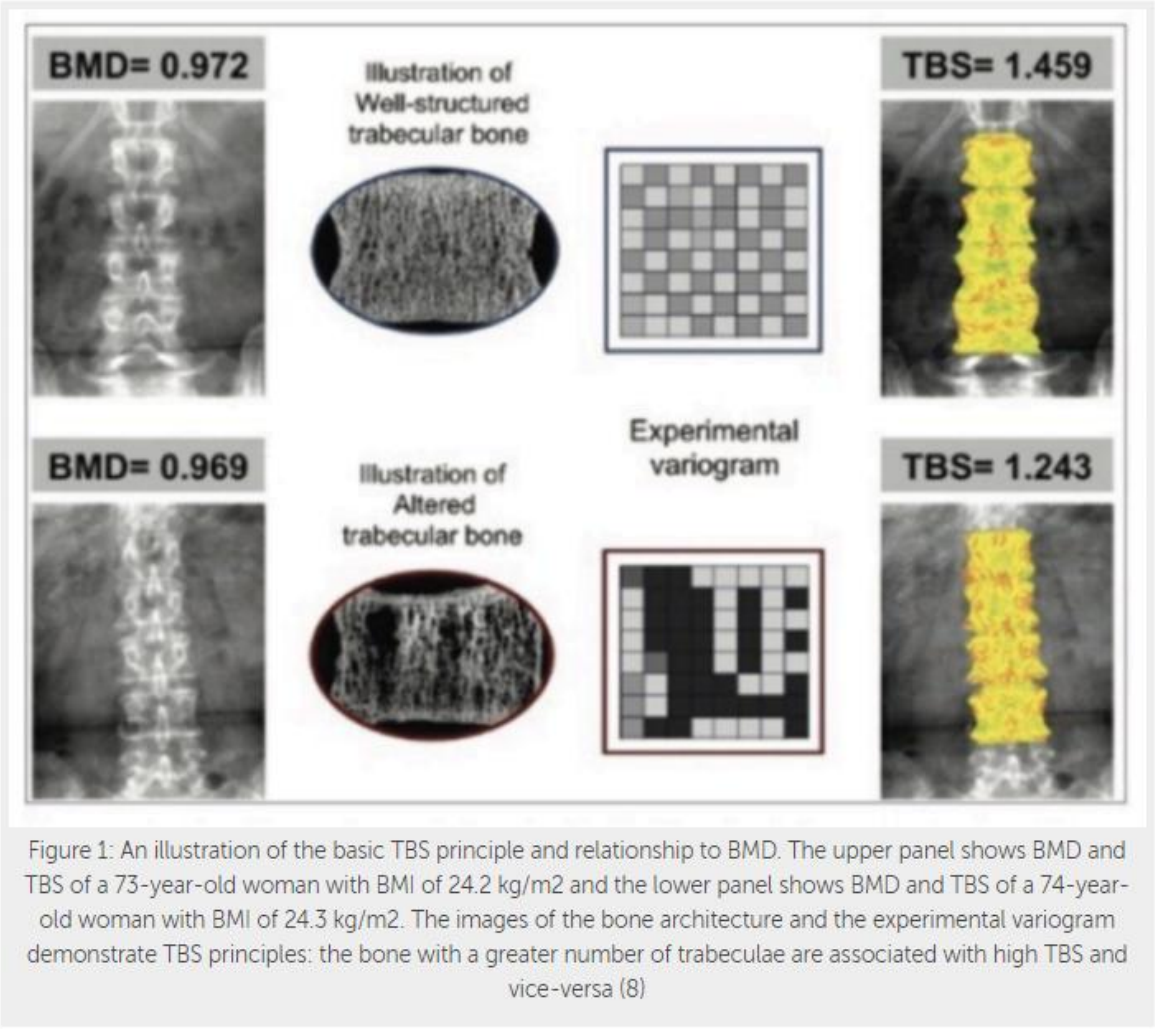


Figure 1: An illustration of the basic TBS principle and relationship to BMD. The upper panel shows BMD and TBS of a 73-year-old woman with BMI of 24.2 kg/m2 and the lower panel shows BMD and TBS of a 74-year-old woman with BMI of 24.3 kg/m2. The images of the bone architecture and the experimental variogram demonstrate TBS principles: the bone with a greater number of trabeculae are associated with high TBS and vice-versa (8)

- Adjunct to aBMD/BMAD in milder OI phenotypes or when aBMD is not osteoporotic
- Caveats are the impact of prior bisphosphonate exposure, spinal rods

Source: Rajan et al. Indian Journal of Endocrinology and Metabolism. 2020; 24:237-243.

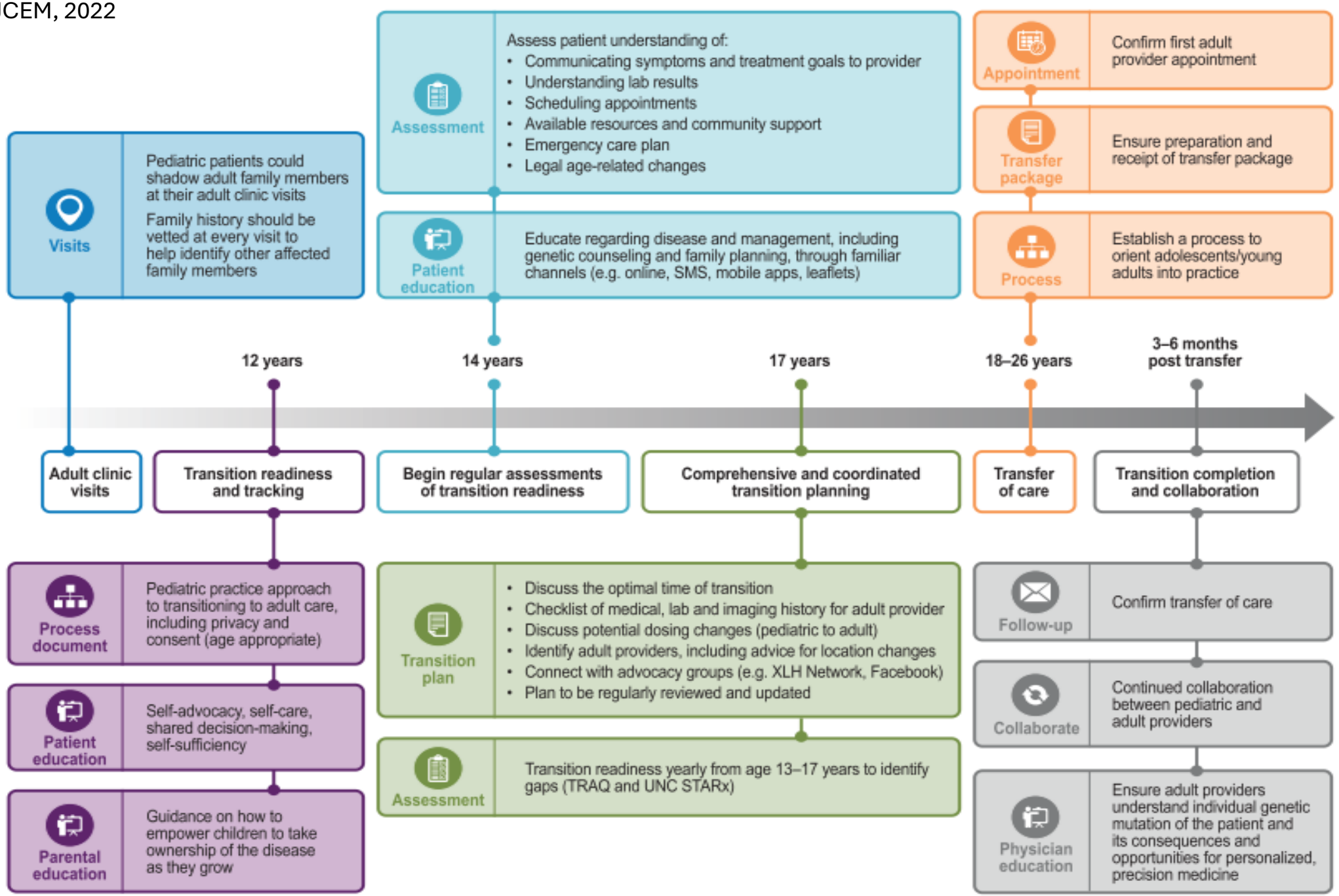
Bone turnover markers in OI

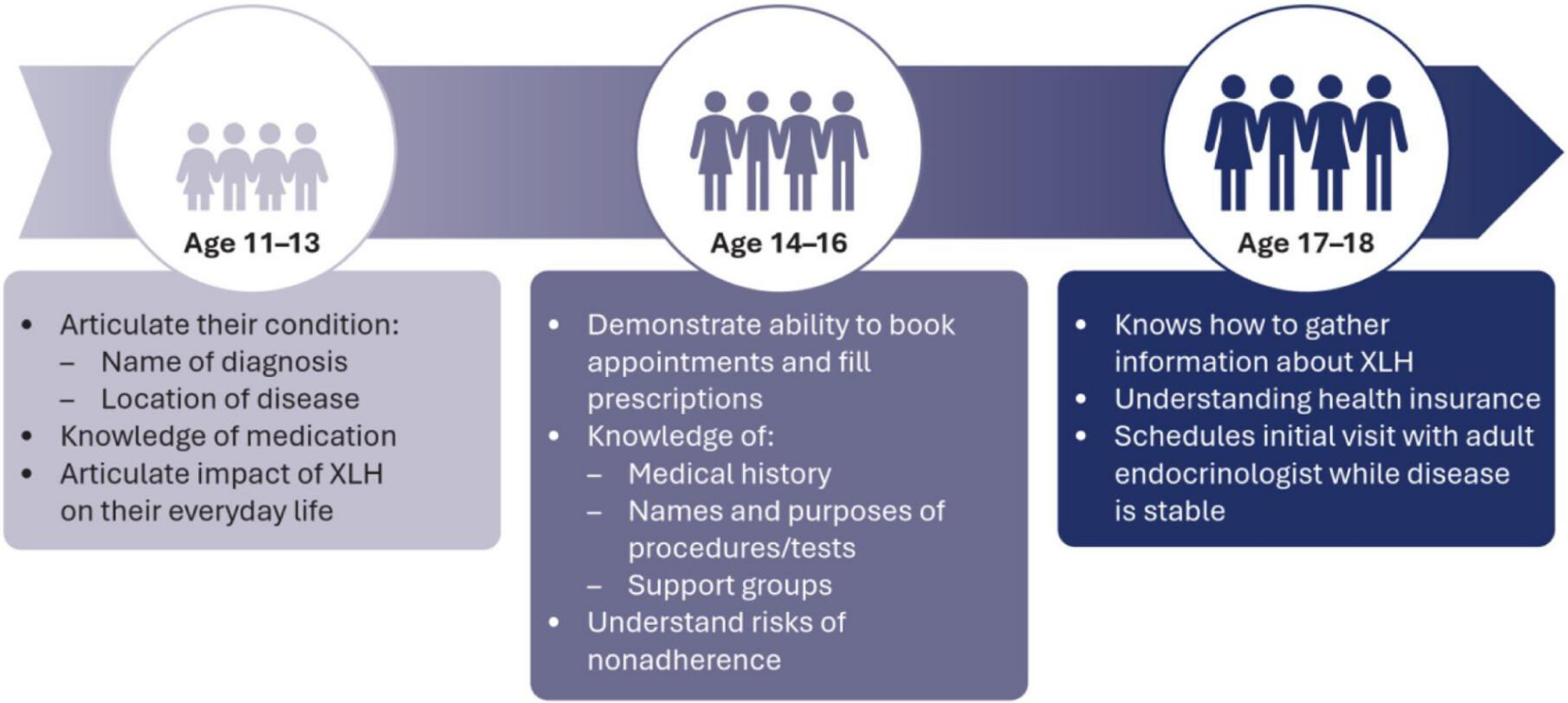
- Bone resorption markers rise after stopping bisphosphonates but stay suppressed compared to the untreated OI population for 2 years
  - CTX more likely to be elevated in children who are still growing

What might a successful bone transition program look like?

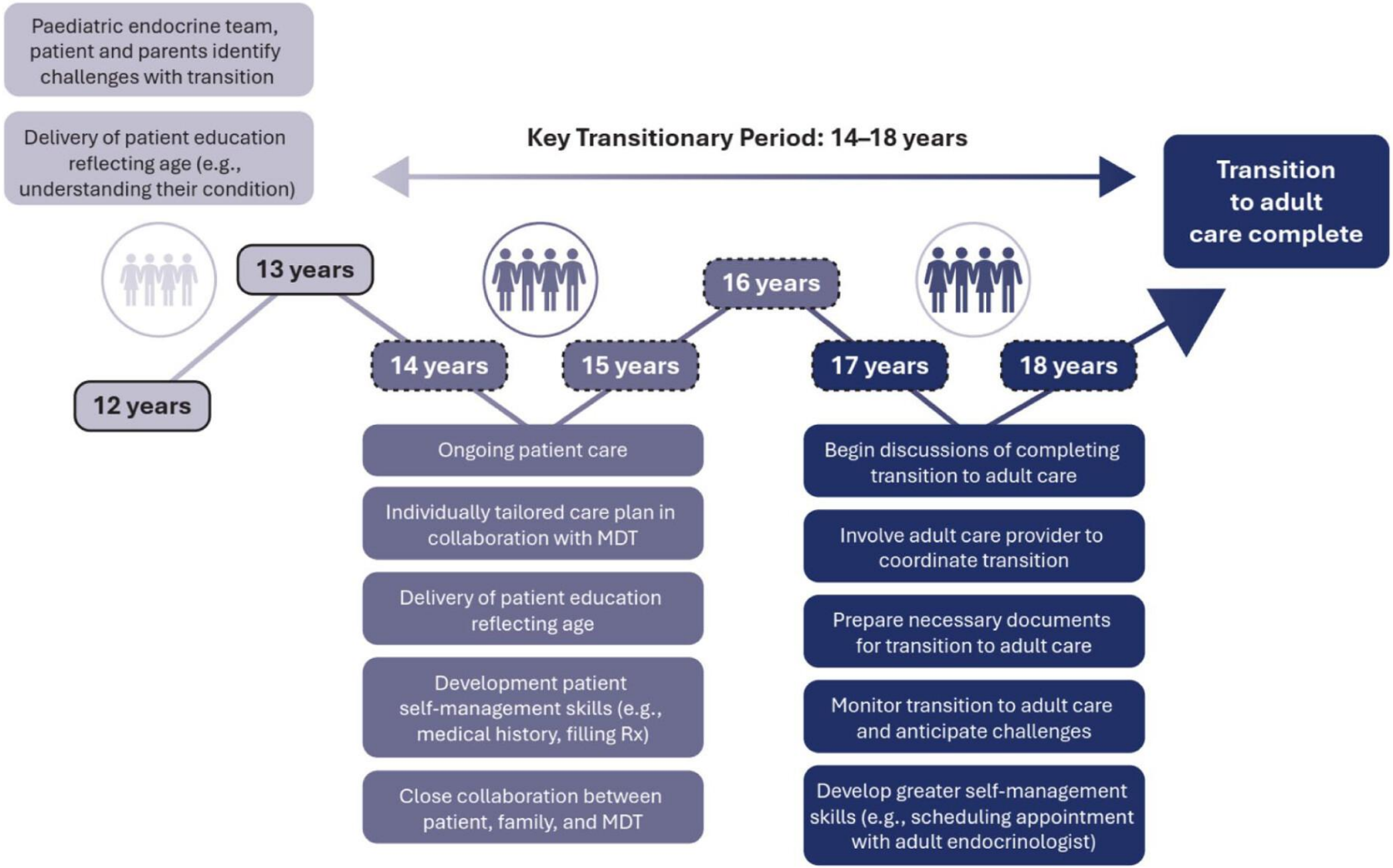
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Dahir et al, JCEM, 2022





Seefried et al *Clin Med Insights Endocrinol Diabetes* 2026

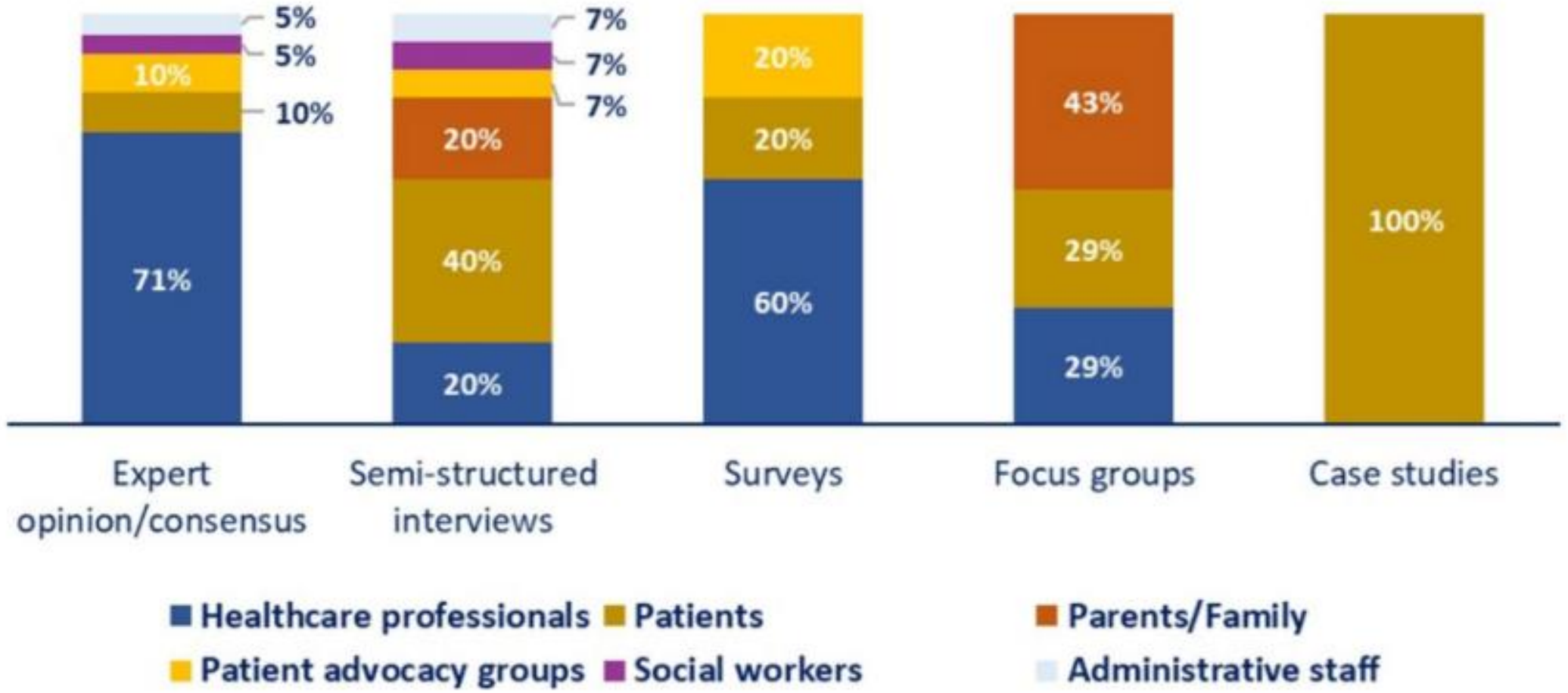


Seefried et al *Clin Med Insights Endocrinol Diabetes* 2026

Tosi et al “Somewhere to go”

- Brought stakeholders to the table in 2023
  - 21 key issues → 3 actionable items to address in the next 3 years
  - All participants carried equal weight/responsibility
- “Center of Excellence” Model
  - Beyond medical expertise
  - Academic/career counseling, financial/legal assistance, transportation services, mental health resources
- Emphasized need for policy reform at a state and federal level

ERN BOND 2026 Scoping Review



Coutinho S et al "Transition Care for Young People with Rare Bone and Mineral Conditions: A Scoping Review." Adv Ther. 2026. PMID: 42313272.

Yaşar 2026 Study

- Assessed the transition of 20 young adults with rare bone disorders
- Multidisciplinary team → written plan providing patient with information on anticipated disease specific challenges in adulthood
- Joint clinics for discussion of reproductive health and mobility issues
- Successful transition (90%) defined as one adult endocrinology visit within 12 months
  - 40% of patients had treatment changes at that visit

What Should Adult Outcomes Look Like

- Historically Measured
  - DXA
  - Fracture rates
  - Lab values
- What Patients Care about
  - Ability to work
  - Mobility
  - Independence
  - Pain
  - Quality of Life

Case Studies

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18yo XLH patient going to college

- Followed approximately every 6 months in endocrine clinic during childhood.
  - Treated with calcitriol and phosphorus but was interested in starting Burosumab
  - Previously had seen nephrology for concerns of medullary nephrocalcinosis and was treated with KCl
    - Kidney ultrasound was “near normal”
  - Has right sided SNHL and vertigo
- Transitioned to Burosumab at 18y 4m of age (1mg/kg) at the start of college

Transition Process

Successes

- Transition occurred at a time when patients life was relatively stable
- Warm hand-off to adult endocrinology (physician and nursing communication)
- First appointment with adult endocrinology occurred 7 months after last peds endo appt
- No missed doses or prescription lapses during the transition process

Mishaps

- Gaps in Care
  - Full imaging prior to transfer of care not done
  - Last renal ultrasound was in 2016
- Written summary not provided
- Formal assessment of transition readiness not completed
- Transitioned later than feasible for many institutions

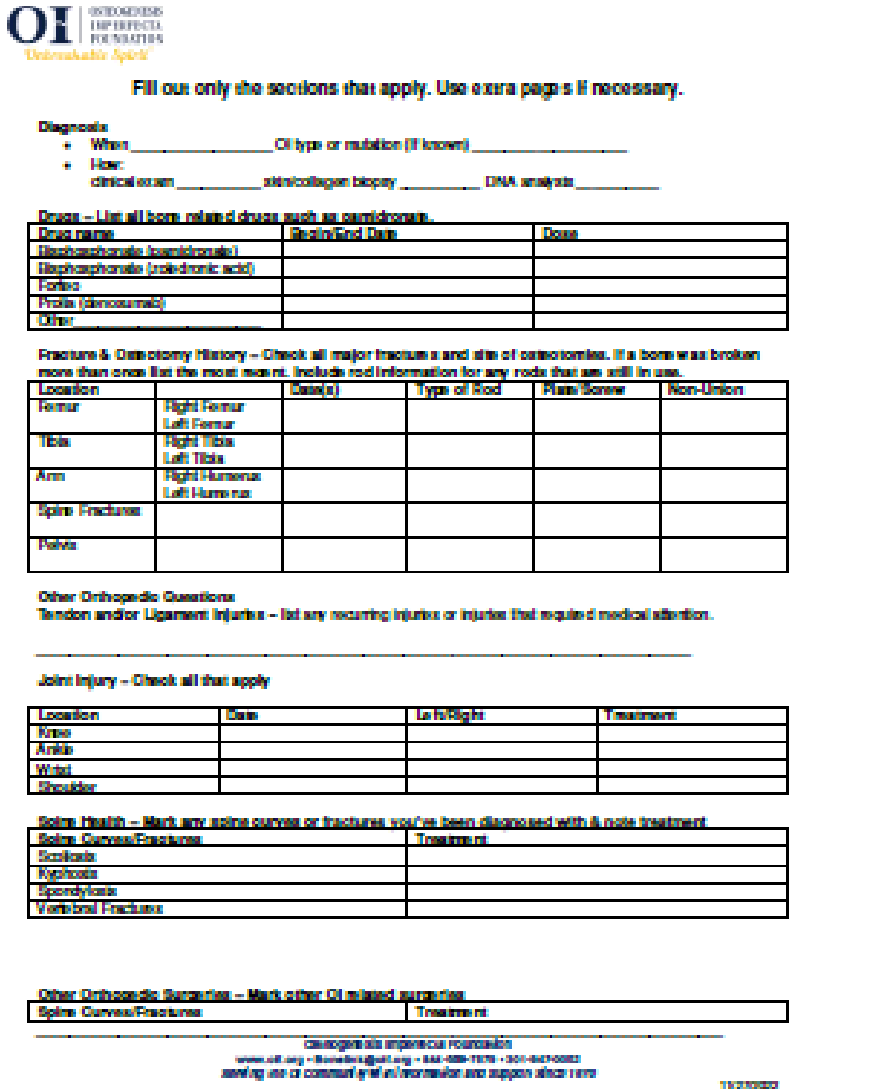
18 yo with OI type 15

- Biallelic variants in *WNT1*
- Over 100 fractures in her birth country of China
- Bisphosphonate infusions given intermittently during early childhood and then with improved regularity at age 8
- Multiple rodding procedures of her lower extremities
- Uses wheelchair for mobility

Challenges with transition planning

1. Identification of skeletal health expert for adult patients with OI
2. Duration of treatment for more severe form of OI – when should a drug holiday be offered?
3. What monitoring should be considered?
    1. DXA site limitations
    2. Bone turnover markers
4. Psychosocial needs assessments
    1. Driving adaptations
    2. College planning
5. Reproductive health considerations

Know Your OI: A Basic Health History



Fill out only the sections that apply. Use extra pages if necessary.

Diagnosis

- When _____ OI type or mutation (if known) _____
- How:
  clinical exam _____ skin/collagen biopsy _____ DNA analysis _____

Drugs – List all bone related drugs such as pamidronate.

Drug name	Start/End Date	Dose
Bisphosphonate (pamidronate)		
Bisphosphonate (zoledronic acid)		
Forteo		
Prolia (denosumab)		
Other _____		

Fracture & Osteotomy History – Check all major fractures and site of osteotomies. If a bone was broken more than once list the most recent. Include rod information for any rods that are still in use.

Location		Date(s)	Type of Rod	Plate/Screw	Non-Union
Femur	Right Femur Left Femur				
Tibia	Right Tibia Left Tibia				
Arm	Right Humerus Left Humerus				
Spine Fractures					
Pelvis					

Other Orthopedic Questions

Tendon and/or Ligament Injuries – list any recurring injuries or injuries that required medical attention.

_____

Joint Injury – Check all that apply

Location	Date	Left/Right	Treatment
Knee			
Ankle			
Wrist			
Shoulder			

Spine Health – Mark any spine curves or fractures you've been diagnosed with & note treatment

Spine Curves/Fractures	Treatment
Scoliosis	
Kyphosis	
Spondylosis	
Vertebral Fractures	

Other Orthopedic Surgeries – Mark other OI related surgeries

Spine Curves/Fractures	Treatment

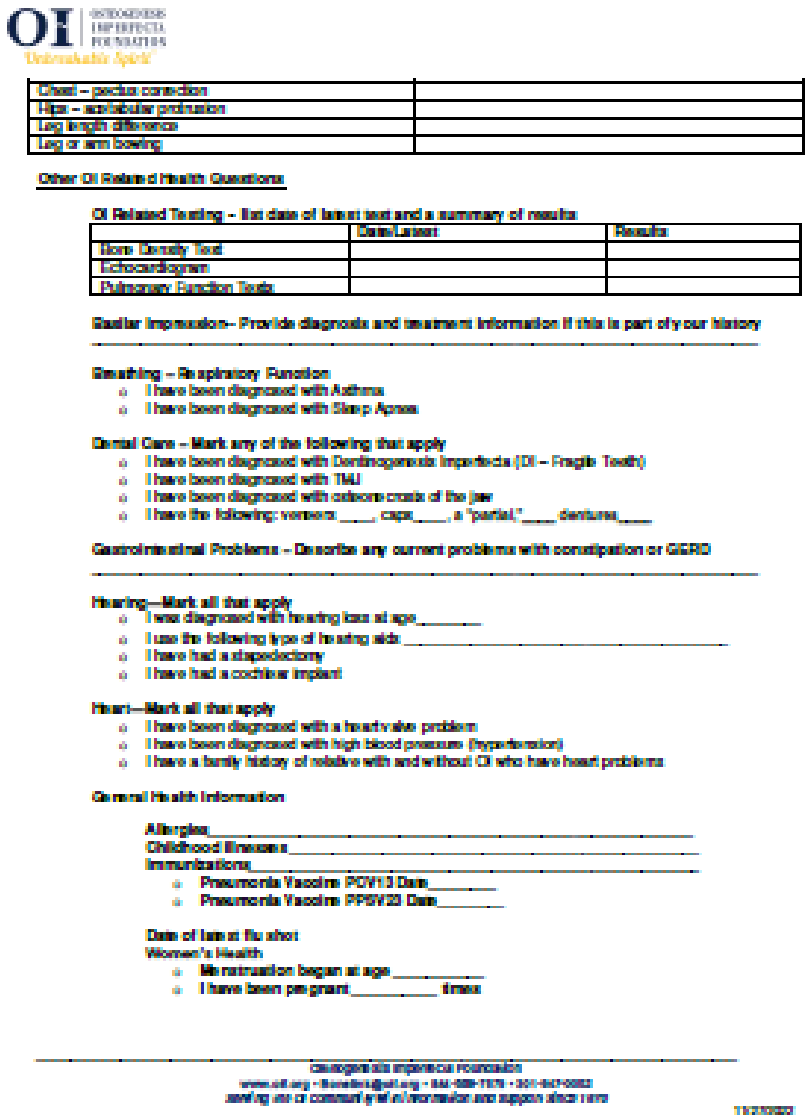
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Osteogenesis Imperfecta Foundation

www.oif.org • Bonebks@oif.org • 844-469-7676 • 301-947-0083

seeking cure & community for all who live with and support those living with OI

10/27/2020



Chest – pectus correction	
Hips – acetabular protrusion	
Leg length difference	
Leg or arm bowing	

Other OI Related Health Questions

OI Related Testing – list date of latest test and a summary of results

	Date/Latest	Results
Bone Density Test		
Echocardiogram		
Pulmonary Function Tests		

Basilar Impression– Provide diagnosis and treatment information if this is part of your history

_____

Breathing – Respiratory Function

- I have been diagnosed with Asthma
- I have been diagnosed with Sleep Apnea

Dental Care – Mark any of the following that apply

- I have been diagnosed with Dentinogenesis Imperfecta (DI – Fragile Teeth)
- I have been diagnosed with TMJ
- I have been diagnosed with osteonecrosis of the jaw
- I have the following: veneers ____, caps____, a "partial,"____ dentures____

Gastrointestinal Problems – Describe any current problems with constipation or GERD

_____

Hearing—Mark all that apply

- I was diagnosed with hearing loss at age_____
- I use the following type of hearing aids _____
- I have had a stapedectomy
- I have had a cochlear implant

Heart—Mark all that apply

- I have been diagnosed with a heart valve problem
- I have been diagnosed with high blood pressure (hypertension)
- I have a family history of relatives with and without OI who have heart problems

General Health Information

Allergies_____
Childhood Illnesses_____
Immunizations_____

- Pneumonia Vaccine PCV13 Date_____
- Pneumonia Vaccine PPSV23 Date_____

Date of latest flu shot

Women's Health

- Menstruation began at age _____
- I have been pregnant_____ times

_____

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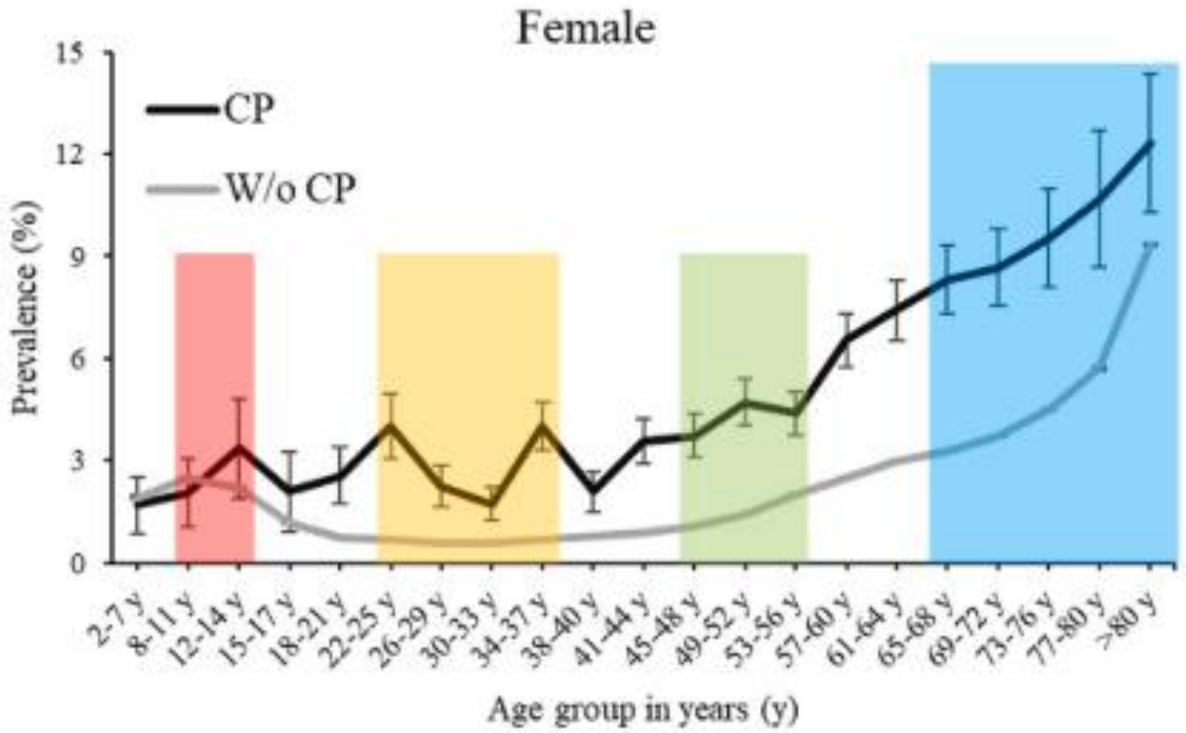
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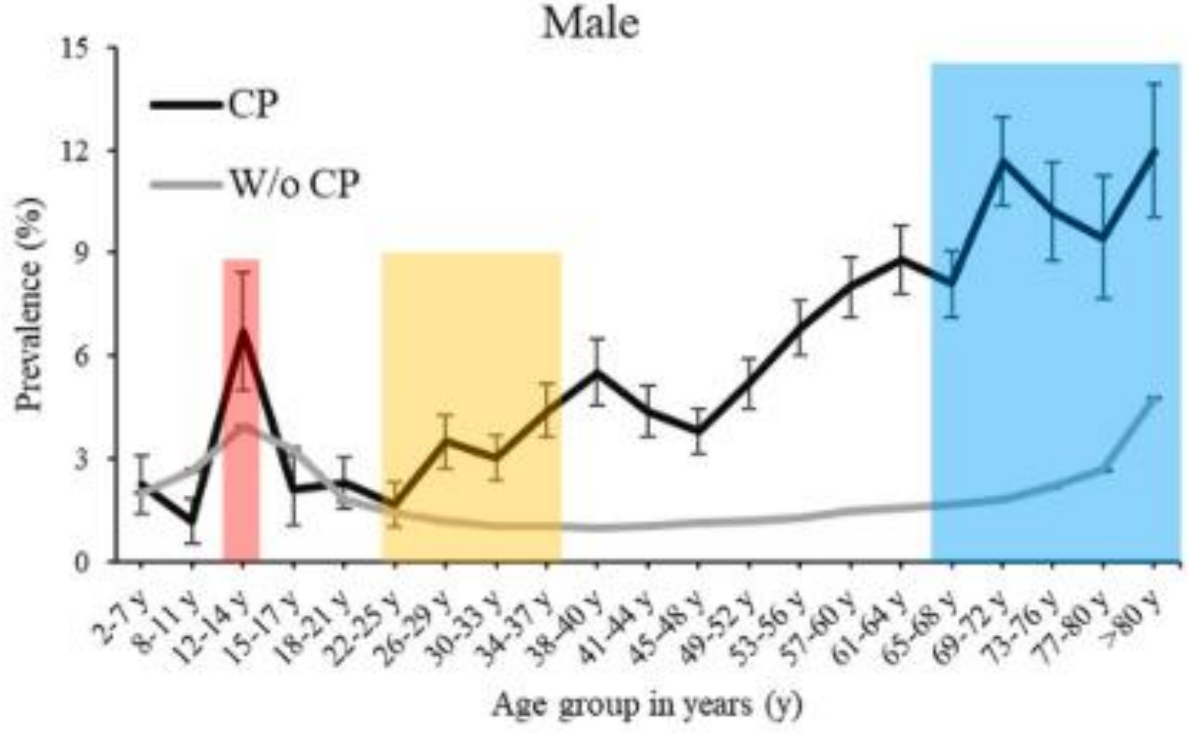
10/27/2020

Secondary Osteoporosis

- Glucocorticoid Induced Osteoporosis
  - Duchenne Muscular Dystrophy
- Disuse Osteoporosis
  - Cerebral Palsy

Fracture Prevalence during Critical Periods of Bone Health





Whitney DG et al. Bone. 2021

SYSTEMATIC REVIEW

Prevalence of and risk factors for osteoporosis and fragility fracture in adults with cerebral palsy: A systematic review

Adult CP Population

- Prevalence of Osteoporosis: 5%
  - 5.5% with history of fragility fracture
- Prevalence of low bone mass by DXA
  - 20-40% at spine and hip
- CP Clinic Population
  - Prevalence of low bone mass: 43%
  - Prevalence of fragility fracture: 38%

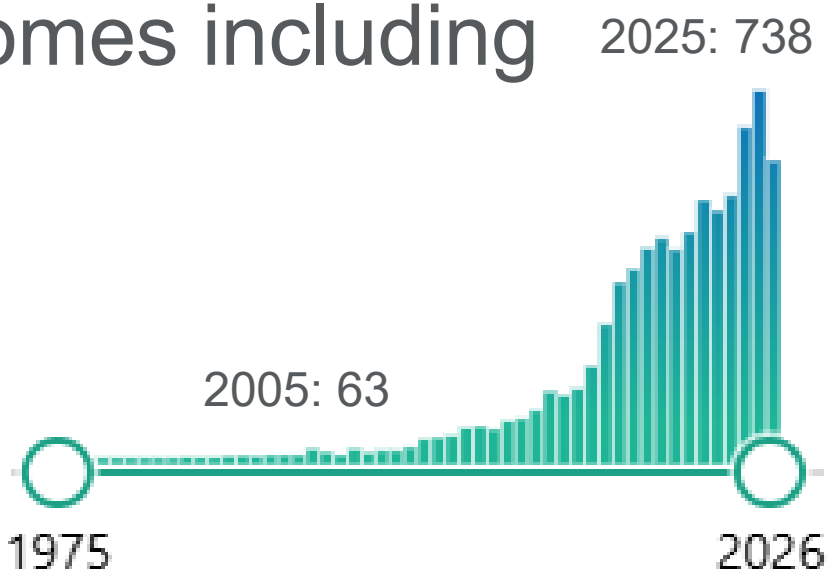
General population ages 50-59 years

- Prevalence of Osteoporosis: 5.1% (USPTF/NHANES)
- Prevalence of fragility fracture
  - Wrist 2.2%
  - Vertebra 0.4%
  - Hip 0.8%

Trinh A et al, Dev Med Child Neurol. 2025

Future Directions – Research Gaps

- Understand the adult burden of disease for skeletal disorders diagnosed in childhood
- Transition metrics: beyond structure and process
- Include quality of life metrics in outcomes including economic impact



Transition of Care Literature: Pubmed

Next Decade of Rare Bone Disease Care

- Artificial Intelligence
  - Fracture prediction
  - Imaging interpretation
- Novel Biomarkers
  - Bone turnover markers
  - Disease activity markers
- Precision Medicine
  - Genotype-guided therapy
- Research Priorities
  - Adult natural history studies
  - Registry-based outcomes
  - Comparative effectiveness studies

Adult Provider Perspective

- Training for congenital rare disease is limited in general
  - Lack of guidelines/evidence for what to do with the young adult patient with skeletal disorders
- Strategies:
  - Disease Centers
  - Shared Care Models
  - National networks (Bone TeleECHO series)

Reciprocal Learning Opportunities

- What can pediatric specialists due to better prepare patients for the adult care system?
- What challenges do adult care providers encounter when receiving patients who were managed for pediatric bone disorders?

References

- Dogba, M. J., Rauch, F., Wong, T., Ruck, J., Glorieux, F. H., & Bedos, C. (2014). From pediatric to adult care: strategic evaluation of a transition program for patients with osteogenesis imperfecta. *BMC health services research*, *14*, 489. https://doi.org/10.1186/s12913-014-0489-1
- Cheung, M., Rylands, A. J., Williams, A., Bailey, K., & Bubbear, J. (2021). Patient-Reported Complications, Symptoms, and Experiences of Living With X-Linked Hypophosphatemia Across the Life-Course. *Journal of the Endocrine Society*, *5*(8), bvab070. https://doi.org/10.1210/jendso/bvab070
- Dahir, K., Dhaliwal, R., Simmons, J., Imel, E. A., Gottesman, G. S., Mahan, J. D., Prakasam, G., Hoch, A. I., Ramesan, P., & Díaz-González de Ferris, M. (2022). Health Care Transition From Pediatric- to Adult-Focused Care in X-linked Hypophosphatemia: Expert Consensus. *The Journal of clinical endocrinology and metabolism*, *107*(3), 599–613. https://doi.org/10.1210/clinem/dgab796
- Westerheim, I., Cormier-Daire, V., Gilbert, S., O'Malley, S., Keen, R., 2024. Osteogenesis Imperfecta: A study of the patient journey in 13 European countries. Orphanet Journal of Rare Diseases 19.. https://doi.org/10.1186/s13023-024-03345-0
- Ross, J., Bowden, M. R., Yu, C., & Diaz-Thomas, A. (2023). Transition of young adults with metabolic bone diseases to adult care. *Frontiers in endocrinology*, *14*, 1137976. https://doi.org/10.3389/fendo.2023.1137976
- Tosi, L. L., Hart, T. S., Beug, G., Carter, E., & Perfetto, E. M. (2025). Somewhere to go: a position paper on addressing gaps in transition care for adults with childhood-onset rare diseases. *Orphanet journal of rare diseases*, *20*(1), 498. https://doi.org/10.1186/s13023-025-03973-0
- Seefried L, Al Badi M, Frookh H, et al. XLH Matters 2024 GCC Edition: Insights and Recommendations to Improve Outcomes for People Living With XLH in the Gulf Region. *Clinical Medicine Insights: Endocrinology and Diabetes*. 2026;19. doi:10.1177/11795514261450032
- Coutinho, S., Jensen, L. S., Gamage, T. H., Hestnes, T. M., Appelmann-Dijkstra, N. M., Bazzocchi, A., Cohen-Solal, M., Crowley, R. K., Eekhoff, E. M. W., Großmann, N., la Forgia, M. C., Travessa, A., Mantovani, G., Masi, L., Micha, D., Oheim, R., Sylvest, T. T., Trisolino, G., Winter, E. M., Horn, J., … Wekre, L. L. (2026). Transition Care for Young People with Rare Bone and Mineral Conditions: A Scoping Review. *Advances in therapy*, *43*(8), 3310–3333. https://doi.org/10.1007/s12325-026-03632-1
- Yaşar, M., Öz Gül, Ö., Nursoy, H., Mercan Sarıdaş, F., Hocaoğlu, E., Karatoprak, K., Denkboy Öngen, Y., Eren, E., Cander, S., Ersoy, C., & Ertürk, E. (2026). Successful Transition in Rare Metabolic Bone Diseases: One-Year Outcomes of a Multidisciplinary Pediatric-Adult Program. *Medicina (Kaunas, Lithuania)*, *62*(7), 1336. https://doi.org/10.3390/medicina62071336
- Collins, L., Lim, A., Alwaheb, S., Chee, N. S. S., Chew, C. K., Farag, G. G. R., Gurdev, K., Zacharin, M., & Simm, P. (2026). Evaluating the transition of young adults with osteogenesis imperfecta: a patient-focused prospective study. *Journal of pediatric endocrinology & metabolism : JPEM*, *39*(8), 768–777. https://doi.org/10.1515/jpem-2025-0273