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Review

Transition of Care in X-Linked Hypophosphatemic Rickets: From Pediatric to Adult Practice- A Narrative Review

Kandemir T et al. XLH Transition: Pediatric to Adult Care

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Abstract
X-linked hypophosphatemic rickets (XLH) requires lifelong, coordinated, and multidisciplinary care, and the transition from pediatric to adult services represents a particularly vulnerable period often accompanied by reduced treatment adherence and a greater risk of loss to follow-up. This review aims to provide a clear, practical framework for supporting the transition of adolescents and young adults with XLH by synthesizing international guidelines, consensus statements, and clinical practice reports published up to August 2025. Current recommendations highlight the importance of early assessment of transition readiness, structured and developmentally appropriate education for patients and their families, close collaboration between pediatric and adult endocrinology teams, and the continuation of therapy with standardized monitoring protocols. A well-designed yet flexible transition pathway may support adherence, ensure continuity of care, and contribute to improved long-term outcomes. By summarizing existing evidence and identifying areas where data remain limited, this review underscores the need for prospective studies to better define optimal management strategies for adults living with XLH.
Keywords: X-linked hypophosphatemic rickets, transition of care, adolescence, adult health, multidisciplinary management, quality of life

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1) Introduction

X-linked hypophosphatemic rickets (XLH) is the most common genetic form of hypophosphatemic rickets, with an estimated prevalence of 1 in 20,000–60,000 and an incidence of 3.9 per 100,000 live births (1,2). Loss-of-function variants in the *PHEX* gene cause increased fibroblast growth factor 23 (FGF23) levels, leading to renal phosphate wasting and impaired vitamin D activation. The resulting disturbances in mineral metabolism give rise to rickets, osteomalacia, dental abnormalities, and skeletal deformities.

Diagnosis is usually established in early childhood, with hallmark features, such as short stature, lower-limb deformities, bone pain, muscle weakness, and recurrent dental abscesses (3–5). In adulthood, additional complications**,** including pseudofractures, osteoarthritis, enthesopathy, hearing loss, and spinal stenosis further impair mobility and reduce quality of life (3,4).

Despite therapeutic advances, including burosumab, long-term outcomes have improved; however, the transition from pediatric to adult care remains a vulnerable phase. This period is often characterized by fragmented care, inconsistent follow-up, and diminished patient engagement (6). According to the American Academy of Pediatrics, the goal of transition is to ensure uninterrupted, developmentally appropriate healthcare that maximizes lifelong function and well-being (7). However, structured transition models remain scarce in rare diseases such as XLH.

Recent regional and international guidelines have highlighted major gaps in patient education, continuity of care, and multidisciplinary coordination during this transition (8–11). These reports emphasize the need for disease-specific transition pathways that are structured, multidisciplinary, and patient-centered. Against this background, the present review consolidates available evidence, synthesizes best practices, and proposes a pragmatic framework for guiding adolescents with XLH into adult care.

2) Methods

This narrative review was conducted through a targeted literature search of PubMed/MEDLINE, Scopus, and Google Scholar to identify articles published between January 2000 and August 2025. The search strategy employed the terms “X-linked hypophosphatemia,” “transition,” “adolescent,” “adult care,” “burosumab,” and “quality of life.” Particular emphasis was placed on clinical practice guidelines, consensus statements, and original investigations that addressed management strategies, transition processes, or long-term outcomes in XLH. In addition, relevant institutional protocols and national policy documents were also reviewed to provide contextual insights.

Given the narrative design, no formal risk-of-bias assessment or meta-analyzes was undertaken. Figures and algorithms included herein represent the authors’ institutional practices refined in alignment with international recommendations. No individual-level patient data were analyzed in this review.

3) Clinical Features of Hypophosphatemic Rickets (XLH)

3a. Childhood and Adolescence

In childhood, XLH is characterized by growth impairment, lower-limb deformities and radiographic features of rickets. Although most infants are born with normal length, growth deceleration typically begins after around six months of age, with short stature often evident by the end of the first year of life (12). Adult short stature is reported in 56–95% of cohorts, even with treatment, reflecting persistent skeletal dysplasia across treatment eras. Disproportion is most often rhizomelic, with relatively short lower limbs and an increased sitting-height ratio.

Lower-extremity deformities, most commonly genu varum or valgum, are highly prevalent. These deformities may delay ambulation, impair gait stability, and contribute to pain and fatigue (13,14). Radiographic abnormalities such as metaphyseal cupping and widening of the long bones are characteristic and may persist in a substantial proportion of patients despite therapy.

Dental involvement is frequent and may present independently of caries or trauma. Recurrent dental abscesses are well described and may coexist with gingivitis, periodontitis, crowding and premature tooth loss, reflecting underlying mineralization defects of dentine and cementum (15).

Additional skeletal findings include widened wrists, rachitic rosary, rib flaring, pectus carinatum and Harrison’s groove. Craniofacial manifestations such as frontal bossing, scaphocephaly, a square-shaped head and craniosynostosis are also recognized (16). Premature suture fusion may result in raised intracranial pressure, particularly in early childhood. Chiari type I malformation occurs in approximately 10% of patients; although often asymptomatic, it may present with headaches or neurological deficits and occasionally necessitate neurosurgical intervention (17).

3b. Adulthood

During late adolescence and adulthood, particularly around the transition period, patients may develop or accumulate complications including pseudofractures, insufficiency fractures, osteomalacia, and progressive loss of mobility (18). Age-associated conditions such as hearing loss, osteoarthritis, enthesopathy, and spinal stenosis are reported with increasing frequency as patients age. Syringomyelia may also occur as a sequela of Chiari I malformation and should be considered in the presence of neurological symptoms.

Prevalence estimates vary considerably across studies and treatment eras, partly due to differences in ascertainment and definitions. Reported ranges include: pseudofractures in ~29–52%, enthesopathy in ~33–100%, and osteoarthritis in ~55–80%. Chronic pain is highly prevalent, and muscle weakness is reported in approximately 60% of adults. Functional limitations, such as reduced six-minute walk distance, are common and correlate with pain, malalignment and degenerative joint disease (19, 20). Dental problems often persist into adulthood, with recurrent abscesses reported in up to 82% of patients. Orthopedic surgery is frequent, with 50–79% of patients undergoing at least one procedure, most commonly long-bone osteotomies (1,20).

Most prevalence estimates derive from cohorts managed with conventional therapy, and the long-term impact of burosumab initiated in childhood on adult outcomes remains to be clarified. Current international consensus emphasizes the need for lifelong surveillance in adults with XLH, with systematic evaluation for fractures, pseudofractures, enthesopathy, dental complications, and hearing loss, given their cumulative impact on quality of life and functional capacity (10).

4) Treatment Approaches

The primary aims of therapy in XLH are to heal rickets, reduce skeletal deformities and the need for surgical, support growth, alleviate pain, preserve dental health, and minimize treatment-related complications.

Conventional therapy consists of multiple daily doses of oral phosphate combined with an active vitamin D analogue (calcitriol or alfacalcidol) (21). This regimen improves clinical, radiographic and dental outcomes in both children and adults. Earlier initiation, particularly within the first year of life, has been associated with more favorable growth, as shown in a United Kingdom cohort where height SDS was significantly better when treatment started before one year (−0.7 vs −2.0, p=0.009) (22).

Doses must be tailored individually. Typical starting regimens include elemental phosphorus 20–60 mg/kg/day in four to six divided doses, calcitriol 20–40 ng/kg/day in two doses, or alfacalcidol 30–50 ng/kg/day once daily. Monitoring should include assessment of growth and clinical symptoms, as well as biochemical surveillance of fasting serum phosphate, calcium, alkaline phosphatase (ALP) and parathyroid hormone (PTH),

together with urinary calcium excretion. Periodic renal ultrasonography is recommended to detect nephrocalcinosis. Careful dose adjustments are essential to avoid overtreatment, which may result in secondary hyperparathyroidism, hypercalciuria, ultimately nephrocalcinosis and renal dysfunction (21).

In adults, therapy focuses on symptoms such as osteomalacia-related pain, muscle weakness, pseudofractures, fractures, and dental complications. Treatment is often indicated around orthopedic or dental interventions, or during pregnancy and lactation. Symptomatic adults may benefit from combined treatment with oral phosphate and active vitamin D analogues, though benefits for enthesopathy and osteoarthritis remain uncertain. Common adult regimens include elemental phosphorus 700–1200 mg/day in two to three divided doses, together with a titrated analogue (most often alfacalcidol). Close biochemical monitoring is required to balance benefits against risks (18). In asymptomatic adults, management decisions should be individualized.

Burosumab, a fully human monoclonal antibody against FGF23, has transformed the therapeutic landscape. By restoring phosphate homeostasis, it promotes skeletal healing and has shown efficacy in both children and adults. Approved by the Food and Drug Administration (FDA) and European Medicines Agency (EMA) since 2018, it is indicated for children from six months of age and for adults with XLH (19,20).

In children, burosumab is administered at 0.8–1 mg/kg every two weeks (up to 2 mg/kg; maximum 90 mg biweekly). In adults, the recommended regimen is 1 mg/kg every four weeks (maximum 90 mg). Dose titration is guided by fasting serum phosphate measured immediately before the next injection, along with ALP, PTH, calcium and urinary calcium. Oral phosphate and active vitamin D analogues should be discontinued prior to burosumab initiation, though adequate native vitamin D should be maintained.

Given its high cost and limited long-term evidence in adults, many centers currently reserve burosumab for patients with more severe disease or for those with inadequate response or intolerance to conventional therapy, in accordance with local reimbursement policies (19–25). The 2025 international working group guideline strongly recommends burosumab over no therapy for adults with fractures or pseudofractures and suggests its use over conventional therapy even in asymptomatic adults, while emphasizing that treatment decisions should remain individualized based on access and comorbidities (10).

During the transition period, ensuring seamless access to therapy and explicit documentation of both dose timing and laboratory sampling relative to treatment are essential to avoid under- or over-treatment and to preserve long-term outcomes.

4a. Orthopedic interventions

Orthopedic surgery is common across the lifespan in XLH, with approximately one-half to four-fifths of patients undergoing at least one procedure. During growth, hemiepiphysiodesis is the typical guided-growth intervention, reported in 14–19% of cases. After skeletal maturity, osteotomy becomes more frequent 17–33% after growth; 34–66% in adult cohorts, whereas hemiepiphysiodesis is rarely used in adults (≈6%) (26,27). Surgery may also be required for fractures and for advanced degenerative joint disease, including hip or knee arthroplasty in selected cases (28). Close peri-operative coordination with endocrinology-to optimize mineral status and avoid both under- and overtreatment- and with rehabilitation services, is essential, particularly during the transition period.

4b. Dental health management

Dental morbidity is a key component of XLH care and requires proactive prevention alongside early specialist involvement. Endodontic procedures, particularly root-canal therapy, are frequently needed in both pediatric and adult patients, reflecting underlying mineralization defects of dentine and cementum (15). Regular dental surveillance and prompt management of abscesses are essential to minimize tooth loss and preserve long-term function.

4c. Rehabilitation

Rehabilitation in XLH focuses on optimizing mobility, function and quality of life. Long-term contribution of physiotherapists, occupational therapists and physical medicine specialists is recommended. Reported use of assistive devices (e.g., walkers, wheelchairs) varies widely, ranging from as low as 11.5% to the majority of patients in certain cohorts, reflecting differences in age, disease severity, and healthcare provision (22, 27). Participation in structured physical therapy is also variable (10–57%) but can help alleviate pain, improve gait, and support reintegration to education or work (27).

4d. Neurosurgical interventions

Neurological comorbidities, including Chiari I malformation, craniosynostosis and syringomyelia, may require neurosurgical procedures such as decompression, cranial surgery. Reported intervention rates are 3.3–9% in children and 2.6–6.3% in adults (26, 27). Patients should be monitored for headaches, visual change, neurological deficits, or signs of raised intracranial pressure, with early referral to neurosurgery when indicated.

4e. Hearing impairment

Hearing loss occurs in 5.7–55% of patients, with prevalence influenced by age, method of ascertainment and treatment era (24,26,27). Formal audiological assessment is recommended from around age 8, or earlier if symptomatic, to enable timely intervention and minimise the impact on development and quality of life.

4f. Other specialties

Comprehensive care is multidisciplinary. Survey data from 43 international experts highlighted consensus that follow-up must involve multiple specialties, including genetic counselling, mental health care, renal ultrasound, and regular dental assessments across both pediatric and adult care (29). Clinical geneticists and genetic counsellors contribute to diagnosis and family planning; psychologists support mental health and treatment adherence; dietitians optimize nutrition; and gynecologists address reproductive health, including pregnancy planning and antenatal care (1,24). Rehabilitation teams further promote educational access and workforce participation. Integrated pediatric to adult models of care are key to optimizing skeletal and dental outcomes, physical function and psychosocial well-being. Age-specific follow-up recommendations are summarized in Tables 1–3. As illustrated in Figure 1, a coordinated multidisciplinary team should include endocrinology, orthopedics, dentistry, audiology, psychology, physiotherapy, nephrology, and genetics.

5) Planning and Completion of Transition Programs

5a. Management of the transition from childhood to adulthood

In hypophosphatemic rickets, a structured, developmentally appropriate transition from pediatric to adult services supports sustained disease control. Planning should begin in early adolescence to ensure continuity of care and minimize treatment gaps. The structured phases of this process from pediatric management through transition to adult services are summarized in Figure 2. Key elements include readiness assessment, progressive self-management training, clear delineation of team roles, and assignment of a coordinator to oversee communication and timelines. Transfer should be accompanied by a written medical summary and a scheduled first adult appointment. Successful completion of transition is defined by attendance at the first adult visit, with a follow-up safety-net review within 3–6 months.

5b. International guidelines and recommendations

Several international initiatives have addressed the transition of care in XLH, reflecting both shared principles and region-specific approaches. The Asia–Pacific consensus recommends initiating preparation for transition as early as 12 years of age, with an emphasis on structured education for patients and families, reinforcement of adherence, and gradual development of self-management skills. Transfer is generally expected between 18 and 21 years, but timing should be adapted to psychosocial maturity and the clinical context (30).

The Latin American consensus proposes a three-stage model that begins with assessment of psychosocial readiness and family support, continues with an active preparation phase including early engagement with the adult care team, and culminates in transfer supported by a detailed medical summary. In this model, educational and social aspects are explicitly considered as part of the process, acknowledging the broader challenges of transition in resource-limited settings (31).

Beyond these regional frameworks, Dahir and colleagues published the first disease-specific consensus for XLH in 2022. This statement highlights the lifelong nature of the disorder and stresses the need for repeated education, including genetic counseling and family planning. It recommends comprehensive skeletal and dental evaluations before transfer, and preparation of a structured transition portfolio containing laboratory data, imaging, and treatment history to ensure continuity of care. The consensus also draws attention to the need for direct communication between pediatric and adult providers, with confirmation of follow-up after transfer to prevent loss to care. In contrast to regional statements, which provide general guidance adaptable to multiple conditions, this document adds specific recommendations tailored to the multisystem burden of XLH (32).

More recently, international working groups have developed systematic GRADE-based guidelines for both children and adults with XLH. These documents provide evidence-based recommendations for diagnosis, treatment including burosumab therapy, monitoring, and transition of care. They complement regional frameworks by offering globally applicable standards while acknowledging the need for individualized approaches (11,33).

Taken together, these publications highlight broad consensus on the importance of early planning, phased preparation, patient empowerment, and multidisciplinary collaboration. Regional frameworks (Asia–Pacific and Latin America) provide pragmatic models that account for healthcare resources and social contexts, while the XLH-specific consensus and international guidelines add precision by defining disease-focused strategies. The combined message underscores that successful transition in XLH requires not only a structured and coordinated process but also tailored interventions that address the lifelong and multisystem character of the disorder.

5c. Transition management in our country

In Turkey, national health insurance covers patients until 18 years of age, with possible extension up to 21 years in chronic conditions. Transition planning generally begins between 14 and 16 years and follows a stepwise process, as shown in Figure 3. The initial stage focuses on patient and family education, assessment of disease knowledge, psychosocial context, and review of prior care. This is followed by reinforcement of self-management skills and an introductory meeting with the adult care team, during which practical arrangements are clarified. The final phase involves formal handover, most often conducted through joint pediatric–adult meetings, with short-term follow-up to secure adherence.

Although shaped by the national health system, this staged model reflects international recommendations: early preparation, clear distribution of role between teams, and a gradual transfer supported by continuous education. Such an approach promotes continuity of care and sustained access to multidisciplinary expertise while remaining feasible in routine clinical practice.

6) Quality of life

Patient-reported outcome measures (PROMs) are central to identifying patient needs and guiding the transition pathway. In practice, age-appropriate validated tools are most effective when applied at several points along the process—for example at baseline, during planning, shortly before transfer, and again a few months after transfer. This allows adjustment of both individual care and service delivery.

From around five years of age, functional status can be assessed using the 6-minute walk test, and with treatment-related improvements documented (34,35). At the beginning of the transition pathway, self-management questionnaires provide insight into disease knowledge and daily self-care, while generic health-related quality-of-life tools such as the SF-36, supplemented by brief screening for anxiety and depression, help to identify psychological and social needs (36). During the planning phase, the Transition Readiness Assessment Questionnaire (TRAQ 5.0) offers a structured way to assess preparedness for adult care. Regional guidelines including the Latin American consensus, also recommend the EQ-5D-5L to monitor physical, mental and social domains, to evaluate treatment effects, and to guide personalized planning (31,38).

Before completion of transition, brief patient-satisfaction surveys and program evaluation forms can be used to audit effectiveness of the model and identify areas for improvement. PROMs therefore inform not only patient care but also overall quality assurance in the transition process.

7) Conclusion

Transition of care in XLH is a critical, multi-year process requiring gradual preparation of both adolescents and families. A structured approach involving the multidisciplinary team, and addressing medical, psychosocial and educational needs, is most likely to ensure a safe and effective handover. This review has synthesized current recommendations and illustrated their adaptation to real-world practice. The proposed algorithm and age-related follow-up tables are intended as practical tools for clinicians and families.

Nevertheless, important gaps remain. Data on long-term adult outcomes are limited, and optimal follow-up intervals and integration of newer therapies after transfer are not yet established. Continuous evaluation of transition programs, tracking first adult visit, prevention of complications and patient-reported quality of life, will be critical. Future multicentre collaborations and registry-based studies should help refine transition models across diverse healthcare systems.

Appendices:

Supplement form: Transition Form

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Statement of Ethics

This article is a review and does not contain any studies with human participants or animals performed by any of the authors. Therefore, Institutional Review Board approval and informed consent were not required.

Conflict of Interest Statement

The authors have no conflicts of interest to declare.

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Table 1. Recommended Monitoring and Assessment Strategy in Pediatric Patients with XLH

Assessment Area	Recommended Parameters	Frequency of Evaluation
Clinical	Height/length, weight, BMI, head circumference, growth velocity, blood pressure	Every 1–3 months during the first year, then every 3 months. Blood pressure monitoring starts after the child becomes cooperative with testing.
	Joint, muscle, bone pain, and headaches	
	Intermalleolar and intercondylar distances, head shape	
Biochemical	Serum calcium, phosphate, creatinine, ALP	Every 1–3 months during the first year, then every 3 months. Creatinine and BUN should be monitored if indicated (e.g., in cases of nephrocalcinosis).
	PTH	
	Urine calcium-to-creatinine ratio	Annually
	25-hydroxyvitamin D	Annually

	1,25-dihydroxyvitamin D	Annually, only in patients receiving Burosumab and depending on resource availability
Radiological	Bone age X-ray	Every 1–2 years or as clinically indicated
	Other skeletal imaging	As clinically indicated
	Renal ultrasound	Annually
Orthopedic	Lower extremity deformities, abnormal gait, or bone pain	Clinical evaluation
Craniofacial	Screening for craniosynostosis, syringomyelia, or Chiari type I malformation	Referral to neurosurgery
Neurosurgical	Monitoring for signs of increased intracranial pressure, such as headache	As clinically indicated
Dental Health	Dental screening and treatment (e.g., periodontitis, dental abscesses)	Twice yearly
Hearing	Audiological evaluation	Begin by age 8, or earlier if indicated. Refer to ENT if hearing loss is suspected.
Genetic Counselling	During diagnosis or family planning	At the time of diagnosis
Functional	Developmental milestones, school participation	Every clinic visit
	6-minute walk test, sit-to-stand test	Annually
Psychosocial	Evaluation via questionnaires	Annually

Abbreviation: XLH: X-linked hypophosphatemia, ALP: Alkaline phosphatase; BMI: Body mass index; BUN: Blood urea nitrogen, PTH: Parathyroid hormone; ENT: Ear-nose-throat.

Table 2. Recommended Monitoring and Assessment Strategy in Adolescent Patients with XLH

Assessment Area	Recommended Parameters	Frequency of Evaluation
Clinical	Height, growth velocity, weight, BMI, blood pressure	Every 1–3 months during the first year, then every 3 months
	Joint, muscle, and bone pain, joint mobility, headaches	
	Intermalleolar and intercondylar distances	
Biochemical	Serum calcium, phosphate, creatinine,ALP	Every 3 months
	PTH	
	Urine calcium-to-creatinine ratio	Annually
	25-hydroxyvitamin D	Annually
	1,25-dihydroxyvitamin D	Annually (only in burosumab-treated patients)
Radiological	Bone age X-ray	Every 1–2 years or as clinically indicated
	Other skeletal imaging	As indicated
	Renal ultrasound	Annually
Orthopedic	Lower limb deformities, abnormal gait, or bone pain in adolescents	Clinical evaluation
Craniofacial	Screening for symptoms of increased intracranial pressure, such as headache	Referral to neurosurgery as needed
Neurosurgical	Monitoring craniofacial complications	As clinically indicated
Dental Health	Dental screening and treatment (e.g., periodontitis, dental abscesses)	Twice yearly
Hearing	Audiological evaluation	Referral to ENT if hearing difficulties are present
Genetic Counselling	For family planning during transition to adolescence	As needed
Functional	School attendance, academic performance	Every clinic visit

	6-minute walk test, sit-to-stand test	Annually, depending on resources
Psychosocial	Assessment using specific questionnaires	Annually, depending on resources

Abbreviations: XLH: X-linked hypophosphatemia; BMI: Body mass index; ALP: Alkaline phosphatase; PTH: Parathyroid hormone; ENT: Ear–nose–throat.

Table 3. Recommended Monitoring and Assessment Strategy in Adult Patients with XLH

Assessment Area	Recommended Parameters	Frequency of Evaluation
Clinical	Height, weight, BMI, blood pressure	Every 1–3 months during the first year, then every 3 months
	Joint, muscle, and bone pain; joint mobility; headaches	
	Intermalleolar and intercondylar distances	
Biochemical	Serum calcium, phosphate, creatinine,ALP	Every 3 months
	PTH	
	Urine calcium-to-creatinine ratio	Annually
	25-hydroxyvitamin D	Annually
	1,25-dihydroxyvitamin D	Annually (only in burosumab-treated patients)
Radiological	Bone age X-ray	Every 1–2 years or as clinically indicated
	Other skeletal imaging	As indicated
	Renal ultrasound	Annually
Orthopedic	Lower limb deformities, abnormal gait, or bone pain in adolescents	Clinical evaluation
Craniofacial	Screening for symptoms of increased intracranial pressure, such as headache	Referral to neurosurgery as needed
Neurosurgical	Monitoring craniofacial complications	As clinically indicated
Dental Health	Dental screening and treatment (e.g., periodontitis, dental abscesses)	Twice yearly
Hearing	Audiological evaluation	Referral to ENT if hearing difficulties are present
Genetic Counselling	For family planning during transition to adolescence	As needed
Functional	6-minute walk test, sit-to-stand test	Every clinic visit
		Annually, depending on resources
Psychosocial	Assessment using specific questionnaires	Annually, depending on resources

Abbreviations: XLH: X-linked hypophosphatemia; BMI: Body mass index; ALP: Alkaline phosphatase; PTH: Parathyroid hormone; ENT: Ear–nose–throat.

Figure 1. Multidisciplinary care team in XLH transition

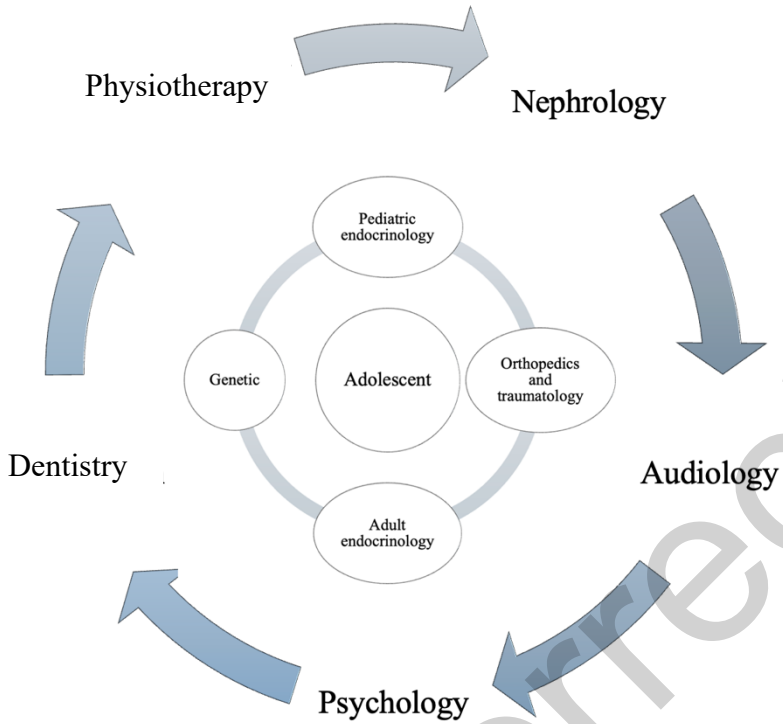


Figure 2. Phases of the transition process in XLH

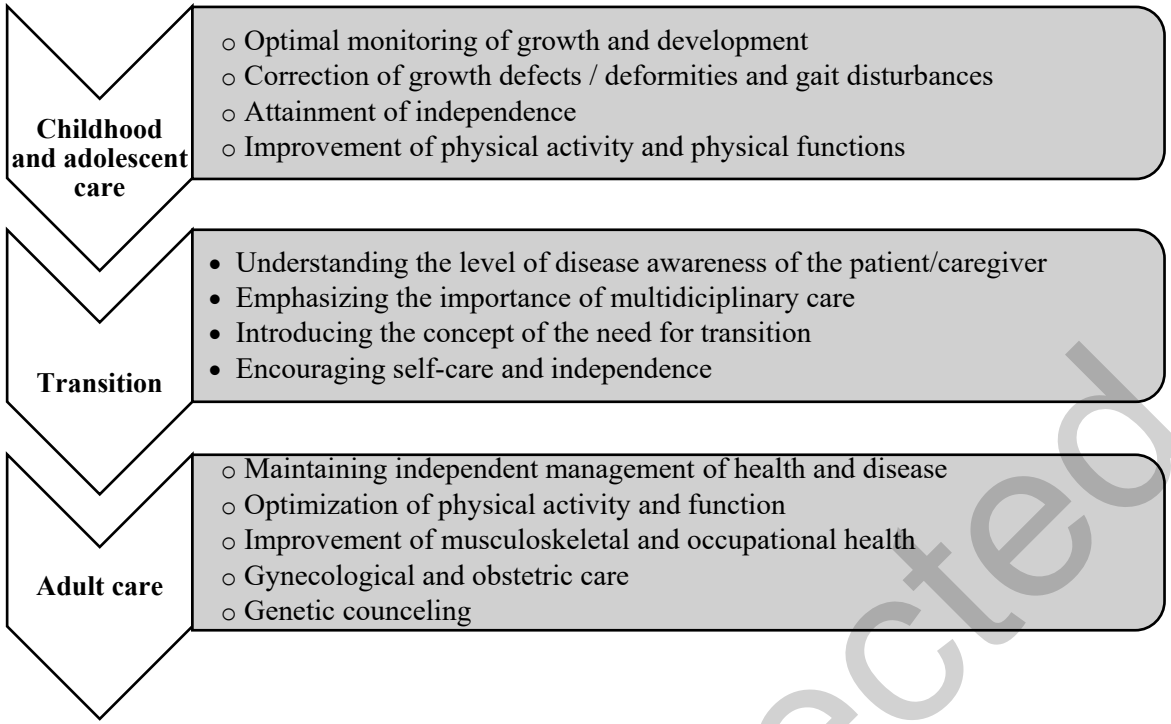
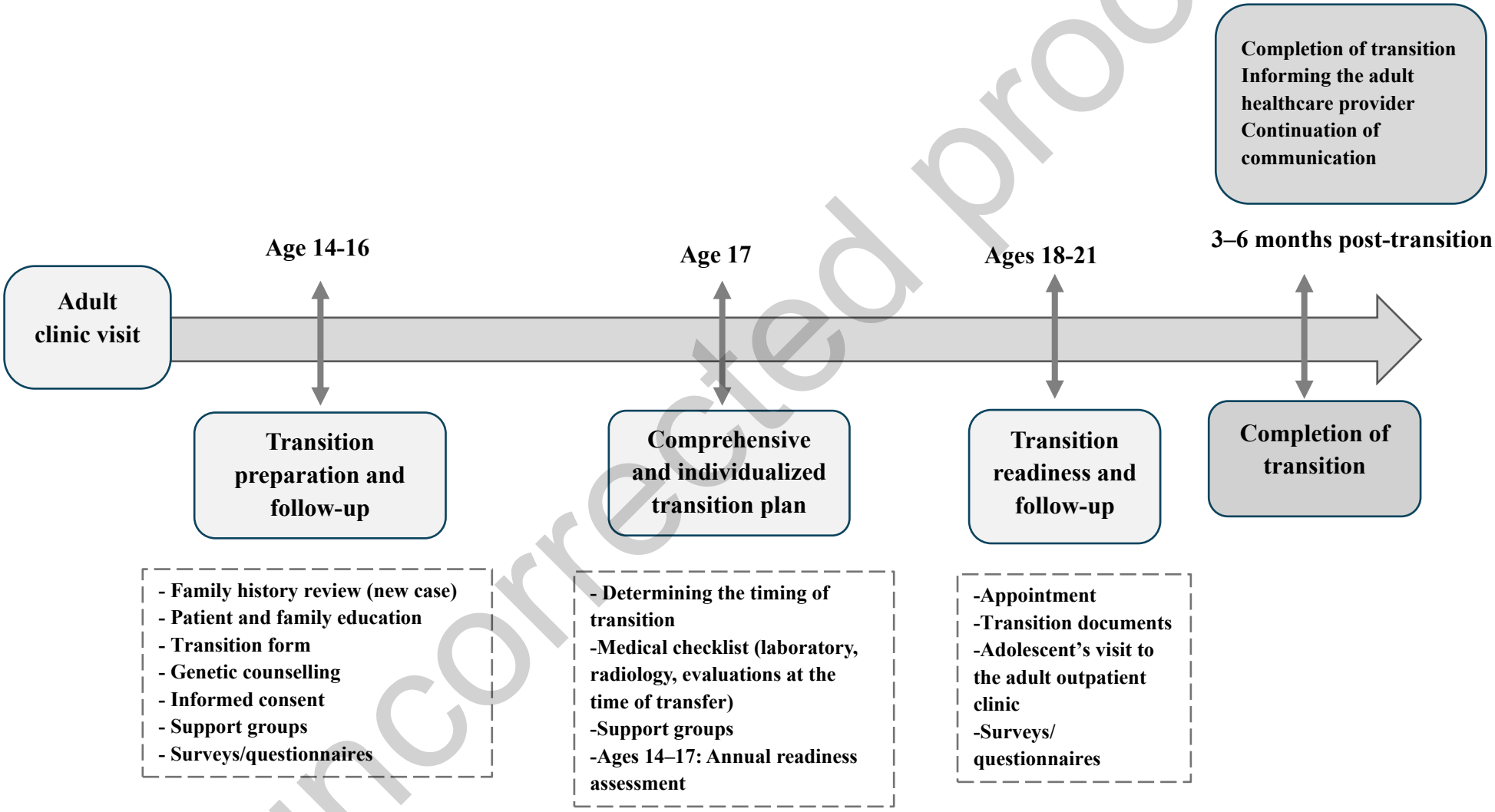


Figure 3. The timeline of the recommended transition process illustrates the stepwise progression from childhood to adulthood



Supplement Transition form
Transition of Care Form in Hypophosphatemic Rickets

Clinical Information		
Full name		
Gender		
Date and place of birth		
Age of diagnosis		
Healthcare institution providing follow-up		
Educational status / occupation		
Date and age at presentation		
Presenting complaint		
Brief medical history at presentation		
Age at last evaluation		
Follow-up intervals / total duration of follow-up		
Family history (consanguinity, similar cases, etc.)		
Maternal height, cm (SDS, percentile)		
Paternal height, cm (SDS, percentile)		
Target height, cm (SDS, percentile)		
Result of genetic analysis		
Physical Examination	**At Presentation**	**At Last Evaluation**
Weight, kg (SDS, percentile)		
Height, cm (SDS, percentile)		
BMI, kg/m² (SDS, percentile)		
Head circumference, cm (SDS, percentile)		
Arm span, cm		
Sitting height, cm		
Sitting height/height ratio		
Intermalleolar distance, cm		
Intercondylar distance, cm		
Deformity		
Systemic findings		
Laboratory Findings	**At Presentation**	**At last evaluation**
Serum Ca / P / Creatinine / GFR		
Alkaline phosphatase		

Spot urine Ca / P / Creatinine		
24-hour urine Ca / P / Creatinine		
Tubular phosphate reabsorption (TPR)		
Renal tubular maximum for phosphate reabsorption per GFR (TmP/GFR)		
25-hydroxyvitamin D		
1,25-dihydroxyvitamin D		
Parathyroid hormone (PTH)		
Intact FGF23		
Bone age		

	At Presentation (Date, age) ……	**At Last Evaluation (Date, age) ……**
Imaging		
Radiographs		
Abdominal–urinary ultrasound		
Cranial CT or MRI		
DXA (if performed)		
Functional Tests		
6-minute walk test		
Specific Evaluations		
Orthopedics		
Dentistry		
Neurosurgery		
ENT – Hearing assessment		
Other		
Medical Treatment (Start date, dosage, etc.)		
Conventional therapy (phosphate, calcitriol, calcidiol)		
Dosage		
Clinical response		
Adverse effects		
Burosumab *Dosage*		
Clinical response		
Adverse effects		
Surgical treatment		
Physiotherapy		
Treatment complications (e.g., nephrocalcinosis, hyperparathyroidism)		
Additional problems		
Pain		
Fractures		
Pseudofractures		
Tooth loss, abscess		

Deformities / Orthopedic problems		
Hearing loss		
Other		
Hospitalizations		
School / work absenteeism		
Quality of Life Questionnaires, date		
SF-36		
EQ-5D		
Brief Fatigue Inventory		