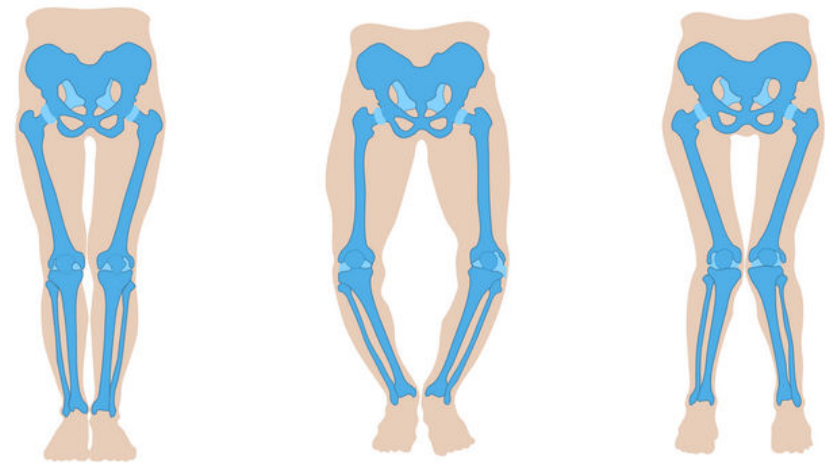


X-linked Hypophosphatemia



Last update: February 24, 2025

Recommended medical check-ups and examinations for XLH

	children	adolescence	adults
frequency of visits	3 to 6 month	3 month	6 to 12 month
height, weight, BMI	✓	✓	✓
knee distance/deformity of the lower limbs	✓	✓	✓
Head circumference/skull shape	✓		
assessment of symptoms such as pain, stiffness and fatigue, and dental status	✓	✓	✓ and detection of signs of osteomalacia, pseudo fractures, osteoarthritis and enthesopathy
assessment of musculoskeletal function (e.g. 6-minute walk test)	annually, as soon as possible	annually	annually
laboratory monitoring: serum levels of alkaline phosphatase, calcium, phosphate, parathyroid hormone, creatinine and GFR	✓	✓	✓
serum phosphate levels and phosphate and creatinine in urine for calculating TmP/GFR (does not require fasting)	when treated with burosumab; frequency of determination varies depending on whether the patient is in the adjustment phase, undergoing dose adjustment or receiving a consistent dose		
25(OH) vitamin D levels	annually		
1,25(OH) vitamin D levels	every 12 months for patients undergoing treatment with burosumab		
calcium-creatinine ratio in urine	every 3 to 6 months		
blood pressure	twice a year		
echocardiography	in cases of persistently high blood pressure		
ultrasound of the kidneys	every 1 to 2 years		
orthopaedic examination	in cases of lower limb deformities		annually in cases of lower limb deformity
X-rays of the left wrist and/or lower extremities	in case of insufficient clinical response to therapy in children; if the leg axis misalignment does not improve in growing patients; in short stature (bone age determination); before orthopaedic surgery; for persistent bone pain in the pelvis or legs		
dental examination	twice a year after tooth eruption	twice a year	twice a year
panoramic image (X-ray)	Ø	if necessary	if necessary
hearing test	from the age of 8 in case of signs of hearing impairment		
examination of the fundus of the eye and cranial MRI	in case of abnormal skull shape, headache or neurological symptoms	in the case of recurring headaches, declining cognitive performance or neurological symptoms	
quality of life (KIDSCREEN-52 questionnaire)	if available (and feasible for children) every 2 years		

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