PS-13

X-Linked Hypophosphatemic Rickets: A case report of 55-year-old female

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Background

- X-linked hypophosphatemic rickets (XLH) is a rare genetic disorder
  - ➢ PHEX gene mutations
  - ➢ Phosphate wasting, hypophosphatemia, and impaired bone mineralization
- While typically diagnosed in childhood, XLH can remain unrecognized until adulthood, presenting with chronic musculoskeletal symptoms that mimic more common rheumatologic disorders.
- We report a case of a 55-year-old female presenting with long-standing musculoskeletal pain and progressive joint stiffness, ultimately diagnosed with XLH through genetic testing.

Case Presentation

- **A 55-year-old female**
- **Chief complaint**
  - ➢ Three-year history of suboccipital pain and chronic low back pain lasting 10 years
- **Past medical history:**
  - ➢ Premature tooth loss at age 30 and progressive difficulty with stair climbing
- **Family history**
  - ➢ Lower limb bowing in her father and paternal grandmother
- **Physical examination**
  - ➢ Short stature (146 cm)
  - ➢ Restricted passive range of motion in multiple joints (cervical/lumbar spine, shoulders, elbows, hips)
  - ➢ Bilateral lower limb bowing

Table 1. Summary of laboratory findings

Parameter	Result	Reference range
Serum phosphorous (mg/dL)	**▼2.4**	2.5–4.5
Total calcium (mg/dL)	9.4	8.6–10.2
Ionized calcium (mmol/L)	1.27	1.05–1.35
Bone-specific ALP (μg/L)	△62.72	7.3–22.4
25-hydroxy vitamin D (ng/mL)	▽25.4	30–100
1,25-dihydroxy vitamin D (pg/mL)	32.95	30–100
PTH, intact (pg/mL)	51.0	15–65
TmP/GFR	**▼0.670**	0.80–1.35
HLA-B27	Negative	Negative

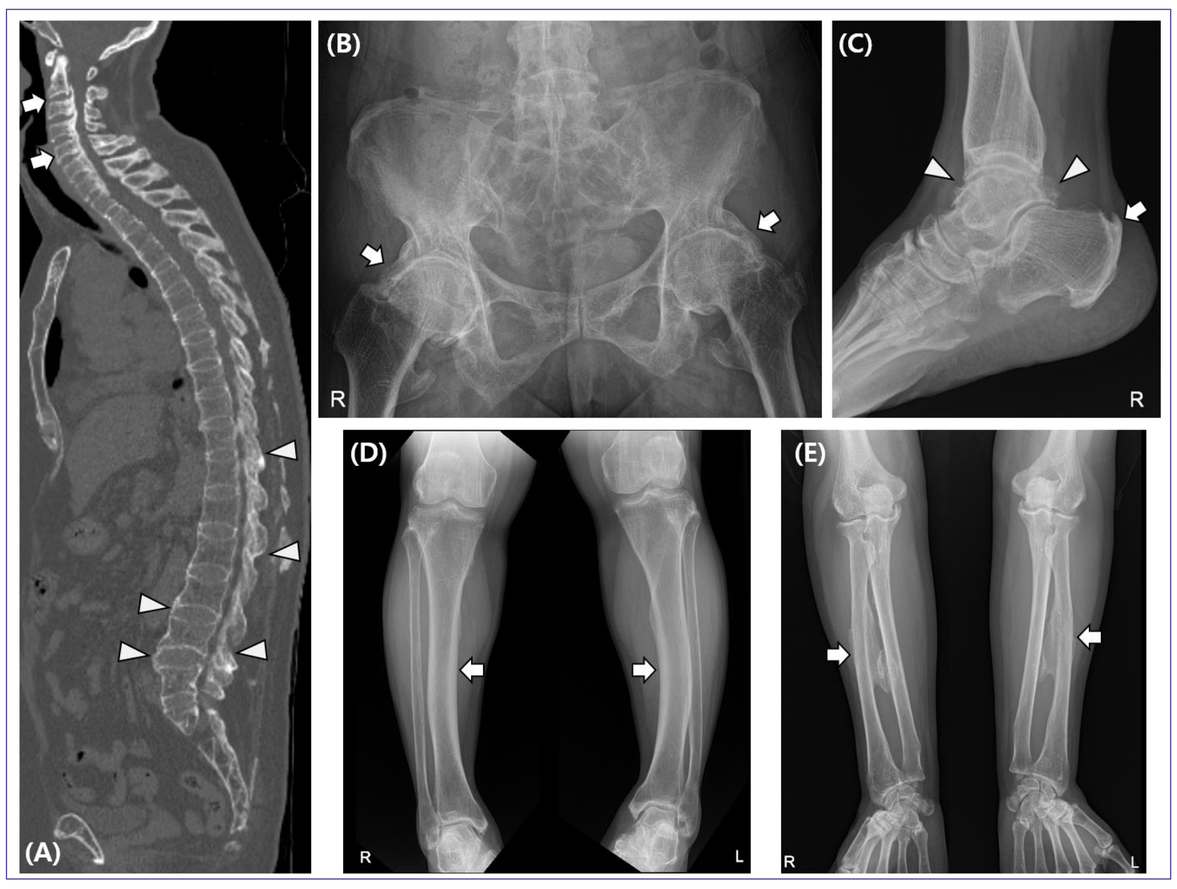

Figure 1. Radiographic and CT findings showing extensive skeletal abnormalities

- ➢ Diffuse syndesmophyte formation, spinal fusion
- ➢ Multiple sites of ossification at tendon and ligament insertion points, indicative of widespread enthesopathy

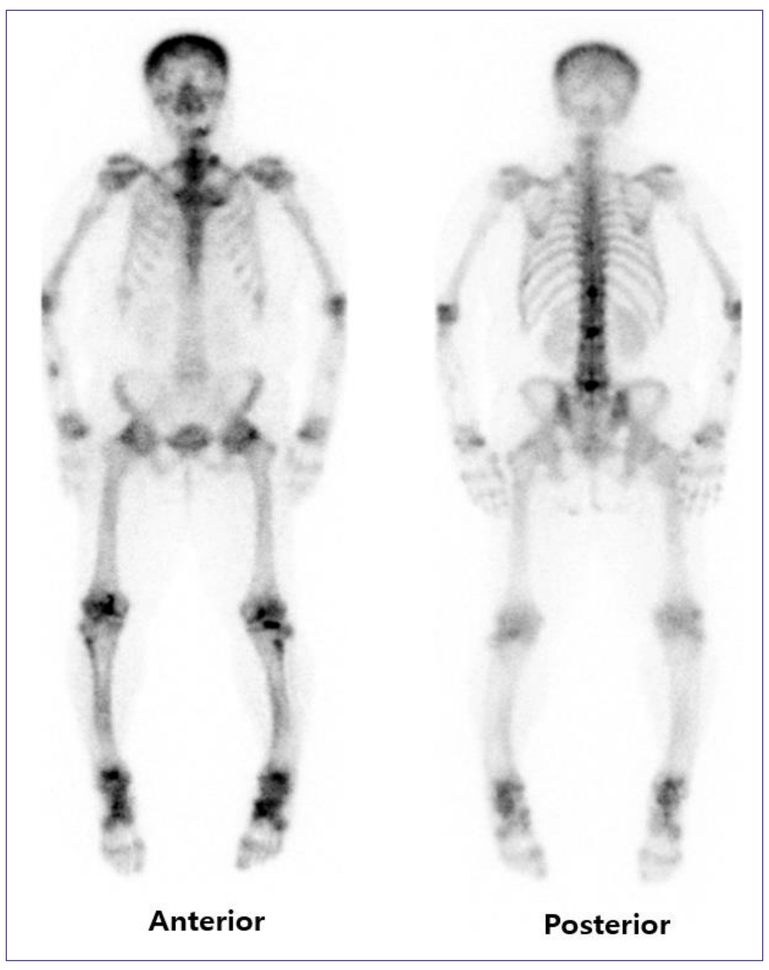

Figure 2. Results of Tc-99m methylene diphosphonate bone scintigraphy

- ➢ Increased radiotracer uptake : T12, L2, and L5 vertebrae, left first rib, right ulna, and both tibia

Diagnosis

- Genetic testing identified a likely pathogenic variant in the PHEX gene (NM_000444.5: c.998_1000del, heterozygous), confirming the diagnosis of XLH rickets.

Treatment

- Oral nonsteroidal anti-inflammatory drugs, phosphate and vitamin D supplementation
- Despite persistent suboptimal phosphorus levels following supplementation, she reported significant pain relief with therapy.

CONCLUSIONS

- ✓ **This case highlights the importance of considering XLH in adult patients with unexplained hypophosphatemia, skeletal abnormalities, and chronic musculoskeletal pain.**
- ✓ **The radiographic findings of enthesopathy and spinal fusion can mimic spondyloarthropathies, potentially leading to diagnostic delay.**
- ✓ **Persistent hypophosphatemia with reduced TmP/GFR and positive family history should prompt genetic testing for XLH, enabling appropriate management of this rare but treatable condition.**