Joint Event on

31st International Conference on

Advanced Clinical Research and Clinical Trials

14th World Congress on

Rare Diseases and Orphan Drugs

June 02-03, 2025

Webinar

A rare case of Vitamin D-dependent rickets Type 1A: Clinical and therapeutic insights

Sagimbayeva Madina*, **Durmanova Aigul**, **Zhaniya Tazhigulova**, **Salykbayeva Zhanat** and **Kurgambekova Diana**
University Medical Center, Kazakhstan

Vitamin D-dependent rickets type 1A (VDDR-1A) is a rare autosomal recessive disorder caused by mutations in the CYP27B1 gene, resulting in impaired conversion of 25-hydroxyvitamin D to its active form. The condition typically presents in early childhood with skeletal deformities, muscle weakness, and growth failure. Here, we present a rare case of a 49-year-old woman with undiagnosed VDDR-1A, illustrating the long-term consequences of delayed recognition and the complexities of late life management.

Case Presentation: A 49-year-old female presented with chronic back pain, progressive muscle weakness, and skeletal deformities, including marked shortening of the lower limbs. She had a long-standing history of physical impairment and was wheel chair dependent.

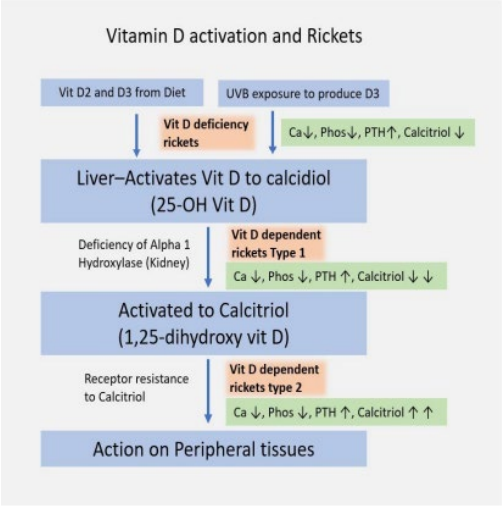
Clinical evaluation revealed: Anthropometric data: height 110 cm, weight 56 kg.

Biochemistry: hypophosphatemia with serum phosphorus 0.71 mmol/L.

Imaging: generalized osteoporosis, multiple fractures, and osteoarthritic changes in the right knee joint. Based on clinical, biochemical, and radiological findings, a diagnosis of VDDR-1A was established. Genetic testing is currently underway to confirm the underlying mutation.

Discussion: This case represents one of the oldest known patients with clinically apparent but previously undiagnosed VDDR-1A. Despite the late diagnosis, initiation of targeted therapy with calcitriol and dietary modifications led to symptomatic improvement. The case emphasizes the potential for positive clinical outcomes even with delayed intervention. It also highlights the need for increased awareness among adult care providers when encountering unexplained skeletal abnormalities or growth disorders.

Conclusion: This case underscores the importance of considering rare inherited metabolic bone diseases such as VDDR-1A, even in adult patients. A multidisciplinary, personalized approach can yield meaningful improvements in quality of life, even decades after symptom onset. Early recognition and timely management remain critical to prevent irreversible skeletal complications.



Biography

Sagimbayeva Madina is a first-year resident in endocrinology, driven by a strong commitment to enhancing patient care and outcomes in hormonal and metabolic health. She is focused on incorporating evidence-based methods into her clinical practice and aims to contribute to the creation of innovative treatments for endocrine disorders. With a solid background in medical education and a keen interest in research, she is dedicated to tackling the challenges associated with diagnosing and managing complex endocrine conditions. Her approach prioritizes patient-centered care, collaboration across disciplines, and a commitment to lifelong learning, all aimed at achieving the highest standards in medical practice.

Received: April 16, 2025; **Accepted**: April 18, 2025; **Published**: June 25, 2025