Following completion of cancer therapy, she was evaluated for parathyroidectomy. Preoperative assessment revealed NYHA class II heart failure, with reduced ejection fraction (36%) and severe tricuspid regurgitation attributed to trastuzumab-related cardiomyopathy. Despite optimal medical therapy, she was deemed high-risk for surgery. Cinacalcet failed to achieve sustained calcium control with levels exceeding 3 mmol/L. She was therefore referred for PEA.

Post-procedure, iPTH decreased 80% by Day 5 (54.9–10.6 pmol/L), with sustained normocalcemia (2.25 mmol/L) at 10 days without further need for cinacalcet.

CONCLUSION

This case illustrates that PEA can serve as definitive therapy for PHPT in patients unsuitable for surgery. It provides rapid and sustained biochemical control, while avoiding operative risk, supporting its role in individualized management.

EP_A092

EXPANDING THE SPECTRUM OF VITAMIN D-DEPENDENT RICKET TYPE 2: DOMINANT-NEGATIVE VDR MUTATION AND POSTPUBERTAL CALCIUM ADAPTATION

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INTRODUCTION

Vitamin D-dependent rickets type 2 (VDDR2) is traditionally defined by biallelic vitamin D receptor (VDR) mutations causing resistance to $1,25(OH)_2D$. We describe a case of the classical VDDR2 phenotype with a single VDR mutation and an interesting physiological adaptation.

CASE

A female diagnosed with rickets at age three presented with a femoral fracture, severe short stature, hypocalcemia (2.1 mmol/L; NR 2.35–2.70), hypophosphatemia (0.8 mmol/L; NR 1.5–2.10), and evidence of renal phosphate wasting (TMP/GFR 0.5 mmol/L; NR 1.5–2.4). Biochemistry showed markedly elevated alkaline phosphatase (1,227 IU/L; NR 50–136) and secondary hyperparathyroidism (20.7 pmol/L; NR 0.8–7.8). Despite hypocalcemia, $1,25(OH)_2D$ was significantly elevated (>450 pmol/L; NR 60–150), consistent with VDDR2. There was no initial family history; however, subsequent evaluation of her mother—prompted by the patient's diagnosis—revealed a similar biochemical profile, along with short stature (142 cm) and a history of multiple fractures. Genetic analysis in both individuals identified a heterozygous missense mutation in exon 10 of the VDR gene, affecting the ligand-binding domain, supporting a pattern of dominant inheritance. The patient was treated with cholecalciferol (1,200 IU daily), calcitriol (5–6 µg daily), and calcium carbonate (2,000 mg daily). Although biochemical responsiveness to therapy was evident, poor adherence resulted in suboptimal metabolic control throughout childhood and adolescence, including a second fracture at age 15 and a final adult height of 124 cm. Notably, calcium and phosphate levels progressively normalized after puberty, with sustained biochemical stability despite the patient omitting the treatment.

CONCLUSION

This case provides two important insights. First, it represents the fourth reported case worldwide demonstrating a dominant-negative effect of a VDR gene mutation, whereby a single mutant receptor interferes with wild-type VDR function. Second, it highlights postpubertal calcium adaptation, in which vitamin D–independent intestinal absorption may restore mineral homeostasis, and disease severity can attenuate over time through adaptive physiological mechanisms.

EP_A093

GIANT PARATHYROID ADENOMA WITH DELAYED HUNGRY BONE SYNDROME: A CASE REPORT

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INTRODUCTION

Giant parathyroid adenomas (GPAs), defined as lesions >3.5 g, are rare. Their size, biochemical severity, and compressive features often mimic carcinoma, creating diagnostic and surgical challenges.

CASE

A 33-year-old female was incidentally found to have hypercalcemia during evaluation after her newborn developed severe hypocalcemic seizures requiring NICU admission. Maternal calcium was 2.95 mmol/L with hypophosphatemia. Subsequent reviews showed persistent hypercalcemia (3.15–3.3 mmol/L), hypophosphatemia (0.32–0.51 mmol/L), and markedly elevated intact parathyroid hormone (85–96 pmol/L). She had vitamin D deficiency, very high alkaline phosphatase (1,329 U/L), and progressive