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UNUSUAL NEUROMUSCULAR AND NEUROPSYCHIATRIC PRESENTATION OF PRIMARY HYPERPARATHYROIDISM: A DIAGNOSTIC CHALLENGE REVEALING AN ECTOPIC MEDIASTINAL PARATHYROID ADENOMA

Endocrinology

Dr. B. Pavan Teja Junior Resident 2nd Year, General Medicine, Government Medical College and Maharashtra Post Graduate Institute of Medical Education and Research, Nashik.

Dr. Abhijit Ramdas Rane* Assistant Professor, General Medicine, Government Medical College and Maharashtra Post Graduate Institute of Medical Education and Research, Nashik. *Corresponding Author

ABSTRACT

Primary hyperparathyroidism (PHPT) can present with vague and multisystem complaints, leading to frequent misdiagnosis and delayed treatment. We describe a 40-year-old woman who presented with progressive myopathy, functional decline to wheelchair dependence, recurrent nephrolithiasis, and nonspecific systemic and neuropsychiatric symptoms. Biochemistry revealed severe hyperparathyroidism with markedly elevated PTH, hypercalcemia, hypophosphatemia, and profound vitamin D deficiency, initially suggesting secondary hyperparathyroidism or hypophosphatemic osteomalacia. Persistently rising PTH despite vitamin D correction prompted further evaluation. Sestamibi scanning localized an ectopic mediastinal parathyroid adenoma, confirmed and removed via VATS excision. Postoperatively, PTH, calcium, phosphorus, and vitamin D normalized, with complete clinical recovery and restoration of mobility. This case highlights how PHPT may masquerade as neuromuscular or neuropsychiatric disease and underscores the importance of considering hyperparathyroidism in patients with unexplained systemic symptoms. Early diagnosis and timely surgical intervention are crucial to prevent irreversible musculoskeletal and neurological complications. **Introduction**: Primary hyperparathyroidism (PHPT) is a common endocrine disorder characterized by inappropriate secretion of parathyroid hormone, most often due to a solitary parathyroid adenoma. While classic manifestations include hypercalcemia, nephrolithiasis, and bone pain, the clinical presentation may be highly variable. Many patients exhibit vague, nonspecific systemic complaints, including fatigue, myalgias, proximal muscle weakness, neuropsychiatric symptoms, or subtle cognitive decline, leading to delays in diagnosis and when compounded by severe vitamin D deficiency, the biochemical profile may mimic secondary hyperparathyroidism or metabolic bone diseases such as hypophosphatemic osteomalacia, further obscuring the underlying pathology. Ectopic parathyroid adenomas, particularly those located within the mediastinum, represent a diagnostic challenge due to their atypical location and inconsistent visibility on routine neck imaging. Failure to identify these lesions may result in prolonged morbidity, recurrent renal stone disease, neuromuscular impairment, and long-term skeletal complications. We report a case of a 40-year-old woman who presented with profound myopathy, recurrent nephrolithiasis, and severe biochemical disturbances initially attributed to secondary causes. Persistent hyperparathyroidism despite vitamin D correction prompted further evaluation, ultimately revealing an ectopic mediastinal parathyroid adenoma. Surgical excision via video-assisted thoracoscopic surgery (VATS) led to complete biochemical normalization and full functional recovery. This case underscores the importance of considering PHPT in patients with unexplained neuromuscular or systemic symptoms and highlights the value of early diagnosis and targeted management in preventing irreversible complications.

KEYWORDS

INTRODUCTION

Primary hyperparathyroidism (PHPT) typically presents with hypercalcemia, nephrolithiasis, and musculoskeletal symptoms. However, ectopic parathyroid adenomas, which account for <5% of cases, may produce vague systemic manifestations, profound neuromuscular weakness, or biochemical patterns overlapping with secondary hyperparathyroidism due to vitamin D deficiency. This diagnostic overlap can delay recognition and definitive treatment. Timely localization and surgical excision are crucial to prevent irreversible bone, renal, and neuromuscular complications. We report a case of PHPT due to an ectopic mediastinal adenoma in a middle-aged woman who initially presented with severe weakness, osteomalacic features, and biochemical abnormalities suggestive of secondary hyperparathyroidism.

Case Presentation

A 40-year-old female, Mrs. Shalini Umesh Kale from Nashik, presented on 19 November 2024 with progressive proximal muscle weakness of several months' duration, resulting in wheelchair dependence and inability to ambulate. She denied sensory symptoms but reported marked fatigue and difficulty performing daily activities. Her past history was notable for recurrent ureteric stones, for which she underwent surgery in 2021. Family history was positive for diabetes in her maternal relatives.

On physical examination, she had severe proximal myopathy with difficulty rising from a seated position. No neck swelling was noted. Clinical suspicion included metabolic bone disease, hypophosphatemic osteomalacia, or secondary hyperparathyroidism.

Initial Laboratory Findings (19/11/2024)

Parameter	Result
Creatine phosphokinase (CPK)	137.5 U/L
Serum Calcium	11.59 mg/dL (elevated)
Serum Phosphorus	1.50 mg/dL (low)
Intact PTH	1087.7 pg/mL (markedly elevated)

Follow-Up Evaluation (06/12/2024)

Parameter	Result
HbA1c	6.1% (pre-diabetes)
Creatinine	0.84 mg/dL
Serum Calcium	9.6 mg/dL
Serum Phosphorus	1.8 mg/dL
25-OH Vitamin D	6.5 ng/mL (severe deficiency)
Intact PTH	1598.7 pg/mL (further elevated)

Repeat Assessment (01/02/2025)

Parameter	Result
Serum Calcium	10.4 mg/dL
Serum Phosphorus	2.2 mg/dL
Vitamin D3	27.2 ng/mL (improved)
Intact PTH	1169.1 pg/mL (still markedly elevated)

Imaging And Localization Studies

Ultrasound Neck

Ovoid hypoechoic lesion (7 × 5 × 6 mm) posterior to the upper pole of the right thyroid lobe with internal vascularity

Additional 8 × 6 mm lesion near the thyroid isthmus (TIRADS 2).

Tc-99m Sestamibi Scan (10/02/2025)

Demonstrated ectopic parathyroid adenoma located in the pre-vascular mediastinal region, anterior to the aortic arch.

Surgical Intervention

On 12 March 2025, the patient underwent Video-Assisted Thoracoscopic Surgery (VATS) for excision of the mediastinal ectopic parathyroid adenoma. The surgery was uneventful.

Postoperative Laboratory Trends

21/04/2025

Parameter	Result
Serum Calcium	8.2 mg/dL
Prolactin	116.2 ng/mL

26/05/2025

Parameter	Result
Serum Calcium	8.2 mg/dL
Serum Phosphorus	2.2 mg/dL

Final Follow-Up (08/08/2025)

Parameter	Result
Serum Calcium	10.2 mg/dL
Serum Phosphorus	4.4 mg/dL
Vitamin D3	56.1 ng/mL
Intact PTH	11.9 pg/mL (normalized)
Creatinine	1.19 mg/dL (eGFR ≈ 58.5 mL/min/1.73 m²)

Clinical Outcome

Following surgery, the patient's neuromuscular symptoms improved dramatically. By August 2025, she was walking independently with no functional limitations. Maintenance therapy included:

Tab Shelcal CT 1-1-1 (30 days)

DISCUSSION

Primary hyperparathyroidism (PHPT) typically results from a solitary parathyroid adenoma; however, its presentation can be highly variable, ranging from asymptomatic hypercalcemia to severe skeletal, renal, and neuromuscular manifestations. This patient's presentation with progressive proximal myopathy, recurrent nephrolithiasis, and profound vitamin D deficiency illustrates the diagnostic complexity of PHPT, particularly when the biochemical profile overlaps with secondary hyperparathyroidism or metabolic bone disease.

At initial evaluation, the combination of severe hypophosphatemia, elevated PTH, and undetectable vitamin D levels raised a strong suspicion for secondary hyperparathyroidism or hypophosphatemic osteomalacia. Vitamin D deficiency is known to suppress calcium levels, stimulate compensatory PTH secretion, and mimic biochemical patterns observed in primary hyperparathyroidism. However, in this case, despite adequate vitamin D repletion, PTH levels continued to rise and hypercalcemia became persistent—key clues pointing to an underlying primary pathology. This reinforces that persistently elevated PTH in the setting of improving vitamin D status should prompt evaluation for PHPT, even when clinical features are nonspecific.

An additional diagnostic challenge in this case was the presence of an ectopic mediastinal parathyroid adenoma, identified only after Tc-99m sestamibi scintigraphy. Approximately 1–5% of parathyroid adenomas occur in ectopic mediastinal locations due to aberrant embryologic migration. These adenomas frequently evade detection on routine cervical ultrasonography, delaying definitive management. Their recognition is critical, as persistent or recurrent hyperparathyroidism after neck exploration often results from an unrecognized ectopic gland.

Management of this patient aligned well with recommended guidelines. Preoperative administration of zoledronic acid was utilized to reduce postoperative bone resorption and mitigate hungry bone syndrome, a known complication following removal of longstanding hyperfunctioning parathyroid tissue. Surgical resection via video-assisted thoracoscopic surgery (VATS) provided a minimally invasive and effective approach for mediastinal adenoma excision. VATS is increasingly favored over sternotomy due to reduced morbidity, shorter recovery times, and excellent visualization of deep mediastinal structures.

The patient's postoperative course demonstrated classic biochemical recovery: normalization of PTH, phosphorus correction, stabilization of calcium, and restoration of vitamin D sufficiency. Clinically, she experienced complete reversal of neuromuscular symptoms, highlighting that myopathy associated with PHPT is reversible when treated early. Her history of recurrent renal calculi and long-standing skeletal symptoms underscores the importance of timely diagnosis, as chronic PHPT is associated with irreversible nephrocalcinosis and osteoporosis when not appropriately treated.

This case illustrates several important learning points. First, PHPT must be considered in patients presenting with vague systemic symptoms, neuropsychiatric complaints, or unexplained myopathy, even when vitamin D deficiency is present. Second, rising PTH levels despite vitamin D therapy are strongly suggestive of primary, not secondary, hyperparathyroidism. Third, ectopic parathyroid adenomas require targeted imaging for localization and may necessitate thoracoscopic intervention. Lastly, coordinated multidisciplinary care -endocrinology, endocrine surgery, radiology, and rehabilitation- ensures optimal outcomes in complex presentations of PHPT.

CONCLUSION

This case illustrates the diagnostic complexity of primary hyperparathyroidism when masked by severe vitamin D deficiency and presenting with vague systemic manifestations such as profound myopathy, fatigue, and recurrent nephrolithiasis. Persistently elevated PTH levels despite vitamin D repletion should prompt clinicians to consider an underlying primary pathology. Targeted imaging is essential, particularly to identify ectopic parathyroid tissue, which may otherwise delay definitive treatment.

In this patient, localization of an ectopic mediastinal adenoma and its successful removal via minimally invasive VATS resulted in complete biochemical normalization and full functional recovery. The case underscores the importance of early diagnosis and timely surgical intervention to prevent potentially irreversible neuromuscular, renal, and skeletal complications. Careful perioperative management, including measures to prevent hungry bone syndrome, further supports optimal patient outcomes. Clinicians should maintain a high index of suspicion for PHPT in patients with unexplained neuromuscular symptoms and persistent hyperparathyroidism, even in the context of profound vitamin D deficiency.

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