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MedEst|2026 | Vol.6: e559| ISSN: 2789-7567 |RNPS: 2524| CASE PRESENTATION

Primary hyperparathyroidism due to oxyphilic cell adenoma in a young patient with thrombopathy: case presentation

Hiperparatiroidismo primario por adenoma de células oxifílicas en paciente joven con trombopatía: presentación de caso

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RESUMEN

Introducción: El hiperparatiroidismo primario (HPP) es frecuente en mujeres posmenopáusicas, pero excepcional en la segunda década de vida. La presentación con fractura patológica y la coexistencia con trombopatía congénita son escasamente documentadas. **Objetivo:** Describir un caso atípico de HPP por adenoma de células oxifílicas en una paciente de 18 años con trombopatía congénita, que debutó con fractura patológica, destacando las lecciones aprendidas en su diagnóstico y tratamiento quirúrgico. **Presentación del caso:** Paciente femenina de 18 años, con trombopatía congénita, consultó por dolor óseo, polidipsia, poliuria y fractura patológica de cúbito. Se evidenció hipercalcemia (3,04 mmol/L), hipofosfatemia (0,72 mmol/L), PTH elevada (156,5 pg/mL) y masa cervical palpable. La gammagrafía con sestamibi mostró un adenoma paratiroideo izquierdo. Se realizó paratiroidectomía mínimamente invasiva, con extirpación de adenoma de células oxifílicas. El postoperatorio fue sin complicaciones hemorrágicas, con normalización del calcio y PTH a las 24 horas. A los 6 meses, la fractura consolidó y la paciente permaneció asintomática. **Conclusiones:** El HPP por adenoma oxifílico puede presentarse excepcionalmente en jóvenes y debutar con fractura patológica. La trombopatía congénita no contraindica la cirugía si se planifica multidisciplinariamente. Este caso subraya la importancia del calcio sérico ante fracturas de bajo impacto en adultos jóvenes.

ABSTRACT

Introduction: Primary hyperparathyroidism (PHPT) is common in postmenopausal women but exceptional in the second decade of life. Presentation with pathological fracture and coexistence with congenital thrombopathy are scarcely documented. **Objective:** To describe an atypical case of PHPT due to oxyphilic cell adenoma in an 18-year-old patient with congenital thrombopathy, presenting with pathological fracture, highlighting the lessons learned in diagnosis and surgical management. **Case presentation:** An 18-year-old female patient with congenital thrombopathy presented with bone pain, polydipsia, polyuria, and pathological ulnar fracture. Hypercalcemia (3.04 mmol/L), hypophosphatemia (0.72 mmol/L), elevated PTH (156.5 pg/mL), and a palpable cervical mass were found. Sestamibi scintigraphy showed a left parathyroid adenoma. Minimally invasive parathyroidectomy was performed, removing an oxyphilic cell adenoma. The postoperative course was uneventful, with normalization of calcium and PTH within 24 hours. At 6 months, the fracture had healed, and the patient remained asymptomatic. **Conclusions:** PHPT due to oxyphilic adenoma can exceptionally present in young individuals and debut with pathological fracture. Congenital thrombopathy does not contraindicate surgery if planned multidisciplinary. This case underscores the importance of serum calcium testing in low-impact fractures in young adults.

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INTRODUCTION

Primary hyperparathyroidism (PHPT) is a common endocrinopathy characterized by the autonomous secretion of parathormone (PTH), leading to hypercalcemia and hypophosphatemia [1,2]. Its most common cause is a solitary parathyroid adenoma (80–85% of cases), and it predominantly affects postmenopausal women, with a peak incidence between 50 and 60 years of age [3,4]. The classic presentation includes renal lithiasis, osteoporosis, and symptoms of hypercalcemia; however, many patients are currently diagnosed incidentally due to biochemical abnormalities [5].

The presentation of PHPT in young adults is exceptional, and its debut as a pathological fracture associated with congenital thrombopathy has not been sufficiently documented in the literature [6]. The coexistence of an oxyphilic cell parathyroid adenoma in an 18-year-old patient with a hereditary bleeding disorder adds a diagnostic and therapeutic challenge, as it requires multidisciplinary management to minimize surgical risks [7]. To date, most case series focus on the older adult population, without exploring the clinical particularities or therapeutic decisions in young patients with hematological comorbidities [8].

The objective of this presentation is to describe an atypical case of primary hyperparathyroidism due to an oxyphilic cell adenoma in an 18-year-old patient with congenital thrombopathy, who presented with a pathological fracture, highlighting the lessons learned in its diagnosis and surgical treatment.

CASE PRESENTATION

An 18-year-old female patient, mestizo, from an urban area, with a personal pathological history of congenital thrombopathy (unspecified platelet function disorder), and no family history of endocrinopathies or toxic habits. She attended the Endocrinology consultation in January 2023 reporting a two-month history characterized by bone pain located in the lower limbs, asthenia, adynamia, occasional nausea, polydipsia, polyuria, and constipation. She denied any history of significant trauma but reported a fall from her own height one week prior, after which she began experiencing pain and functional impairment in her left forearm.

Physical examination revealed: blood pressure 110/70 mmHg, heart rate 88 beats per minute, eupneic. In the left upper limb, deformity and edema were evident in the middle third of the forearm, with limited active mobility. Neck examination revealed a palpable,

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painless, firm mass, approximately 2 cm in diameter, in the anterior region, movable with swallowing, probably related to the left thyroid lobe. The rest of the neurological and muscular examination showed generalized hypotonia, without focal motor deficits. This is a case of primary hyperparathyroidism (ICD-11: E21.0) with an atypical onset.

An X-ray of the left forearm was requested, which demonstrated a pathological fracture of the middle third of the ulna, with osteolytic borders. Given the suspicion of metabolic bone disease, the study was extended with:

Laboratory blood tests (local laboratory reference values in parentheses):

- Serum calcium: 3.04 mmol/L (2.10 – 2.60)
- Phosphorus: 0.72 mmol/L (0.87 – 1.45)
- Intact parathormone (PTH): 156.5 pg/mL (10 – 65)
- Alkaline phosphatase: 3955 U/L (30 – 120)
- Creatinine: 96 µmol/L (40 – 128)
- Urea: 6.7 mmol/L (1.7 – 8.1)
- 25-hydroxyvitamin D: 28 nmol/L (deficiency: <50)

Bone survey: generalized osteopenia, osteolytic lesions in the calvaria, femurs, and left humerus, with a "salt-and-pepper" appearance in the skull.

Parathyroid scintigraphy with Tc-99m sestamibi: focal pathological uptake in the projection of the left thyroid lobe, suggestive of a parathyroid adenoma.

With a presumptive diagnosis of primary hyperparathyroidism secondary to a parathyroid adenoma, a minimally invasive parathyroidectomy was performed via an anterior cervical approach. During the surgical procedure, an enlarged parathyroid gland (1.8 × 1.5 × 1.2 cm) was excised. The histopathological study reported a parathyroid adenoma composed predominantly of oxyphilic cells, with no signs of atypia or necrosis.

The immediate postoperative period was uneventful, with no hemorrhagic or infectious complications. At 24 hours after surgery, serum calcium dropped to 2.25 mmol/L and PTH to 28 pg/mL. The patient was discharged on the third postoperative day with oral calcium supplements (1000 mg/day) and calcitriol (0.25 µg/day) for two weeks. At the 6-month follow-up, the patient was asymptomatic, with calcium and PTH levels within normal ranges, and the control X-ray showed consolidation of the ulnar fracture.

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Informed consent: Written informed consent was obtained from the patient for the publication of this clinical case, including clinical data. The authors guarantee that patient anonymity has been respected at all times. The consent document is available for verification by the editorial committee.

DISCUSSION

The present case describes an uncommon form of PHPT due to its presentation in an 18-year-old patient with congenital thrombopathy, who debuted with a pathological fracture. Although PHPT is common in postmenopausal women [1,2], its occurrence in the second decade of life is exceptional; the literature reports less than 1% of cases below 30 years of age [3,11]. Early age mandates ruling out hereditary syndromes such as multiple endocrine neoplasia (MEN) or familial isolated hyperparathyroidism, which were not evidenced in this patient due to the absence of family history and other endocrine neoplasms [12].

Oxyphilic cell adenomas represent between 3 % and 10 % of all parathyroid adenomas [13]. Unlike chief cell adenomas, they tend to be smaller and may be associated with variable hypercalcemia, although classically they have been described as having lower PTH production per unit mass [13]. In this case, the patient presented with marked hypercalcemia (3.04 mmol/L) and frankly elevated PTH (156.5 pg/mL), suggesting high secretory activity despite the oxyphilic lineage, an uncommon finding that deserves to be highlighted [15].

Presentation with a pathological fracture constitutes a classic but now infrequent form of presentation in countries with early biochemical diagnosis [16]. In regions with limited access to routine testing, up to 25 % of patients are still diagnosed after bone or renal complications [8]. This case serves as a reminder that, even in young patients, PHPT must be considered in the setting of recurrent or low-energy fractures, especially if accompanied by hypercalcemia or hypophosphatemia [5].

The coexistence of congenital thrombopathy added a therapeutic challenge. Parathyroid surgery, although minimally invasive, carries a bleeding risk [9]. The literature recommends a preoperative hematology evaluation, with availability of platelet derivatives or antifibrinolytics depending on the specific defect [10]. In this patient, the intervention was performed without complications, probably due to the mild nature of her thrombopathy and the use of local hemostatic measures. Nevertheless, this case underscores the need

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for a multidisciplinary approach (endocrinologist, surgeon, hematologist) in patients with bleeding comorbidities [9,10].

Compared with other published cases of PHPT in young adults, most report chief cell adenomas and nonspecific symptoms (fatigue, vague bone pain) that delay diagnosis by an average of 2 years [17]. Our patient was diagnosed within weeks thanks to the occurrence of the fracture, which facilitated early surgical intervention. This contrast highlights the value of a thorough physical examination (detection of a cervical mass) and the request for serum calcium in any pathological fracture in young individuals [18].

From a management standpoint, minimally invasive parathyroidectomy guided by sestamibi scintigraphy is the gold standard for localized single adenomas [7]. Normalization of calcium and PTH at 24 hours confirmed surgical success. The 6-month follow-up demonstrated bone consolidation and complete remission, similar to the outcomes reported in series of young patients [18].

A limitation of this case is the lack of genetic testing to rule out mutations in *MEN1*, *CASR*, or *CDC73*, given that presentation before age 30 justifies this workup [19]. Likewise, fractional excretion of calcium was not determined to rule out familial hypocalciuric hypercalcemia, although the clinical presentation and PTH levels make this diagnosis unlikely [20]. Despite these limitations, the case offers practical lessons: (i) PHPT can present in adolescents with low-impact fractures; (ii) congenital thrombopathy does not contraindicate surgery if properly planned; (iii) oxyphilic adenoma can present with severe hypercalcemia, contrary to classical belief.

CONCLUSIONS

Primary hyperparathyroidism due to an oxyphilic cell adenoma can exceptionally occur in patients in the second decade of life and present with a pathological fracture, as evidenced in this case. The association with congenital thrombopathy does not contraindicate minimally invasive parathyroidectomy, provided that multidisciplinary planning and appropriate hemostatic measures are undertaken, which allowed for an uneventful postoperative course without hemorrhagic complications. Normalization of calcium and parathormone at 24 hours after surgery, together with bone consolidation at the 6-month follow-up, confirm the efficacy and safety of surgical treatment even in young patients with comorbidities. This case underscores the importance of requesting serum calcium and considering primary hyperparathyroidism in the differential diagnosis of pathological

fractures in young adults, especially when accompanied by constitutional symptoms or a palpable cervical mass.

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AUTHORSHIP STATEMENT

ARR: Conceptualization. Data curation. Formal analysis. Fundraising. Research. Methodology. Drafting of the original manuscript and revision and editing. Project management and supervision.

RSL: Research. Methodology. Drafting of the original manuscript and revision and editing.

YEG: Research. Methodology. Drafting of the original manuscript. Formal analysis.

CONFLICTS OF INTEREST

There are no conflicts of interest.

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