Primary hyperparathyroidism associated with genu valgum in a young girl: Report of a rare case

BAYAR A. QASIM[1], LAWEEN O. MUSA[2], AYOOB M.T. ABDULRAHMAN[3], SOHAIB H. ALI[1], HALDER J. ABOZAIT[4] and ABDULKAREEM M. JOMAA[1]

[1]Department of Medicine, College of Medicine, University of Duhok, Duhok, Kurdistan Region 42001, Iraq; [2]Department of Surgery, College of Medicine, University of Duhok, Duhok, Kurdistan Region 42001, Iraq; [3]Heevi Paediatric Teaching Hospital, Duhok, Kurdistan Region 42001, Iraq; [4]Department of Biomedical Sciences, College of Medicine, University of Zakho, Zakho Independent Administration, Kurdistan Region 42002, Iraq

Received March 10, 2026; Accepted July 10, 2026

DOI: 10.3892/mi.2026.332

Abstract. Primary hyperparathyroidism (PHPT) is rare in children and may present with skeletal deformities. The present case report describes the case of a 12-year-old girl with progressive bilateral genu valgum and bone pain unresponsive to corrective osteotomy and supplementation. She had a prior episode of hypercalcemia during acute pancreatitis. Laboratory analyses revealed elevated calcium levels (12.2 mg/dl), markedly high levels of parathyroid hormone (PTH; 1,574 pg/ml), low vitamin D levels (13.3 ng/ml) and increased levels of alkaline phosphatase (2616 U/l). A neck ultrasonography revealed a right parathyroid adenoma, and radiographs revealed multiple expansile, cystic lesions with a 'soap-bubble' appearance. A histopathological analysis confirmed osteitis fibrosa cystica (brown tumor). Radiofrequency ablation of the adenoma normalized the calcium and PTH levels, with the near-complete radiological resolution of lesions and the marked clinical improvement of genu valgum. The present case report highlights a rare pediatric presentation of PHPT with osteitis fibrosa cystica and genu valgum, initially misdiagnosed as an orthopedic disorder. Early endocrine evaluation in children with unexplained bone deformities is essential to prevent unnecessary surgical interventions and morbidity. Successful management of the parathyroid lesion led to substantial, although not formally quantified, clinical and radiological improvement.

Correspondence to: Dr Halder J. Abozait, Department of Biomedical Sciences, College of Medicine, University of Zakho, Zakho International Road, Zakho Independent Administration, Kurdistan Region 42002, Iraq
E-mail: halder01jamal@gmail.com

Key words: primary hyperparathyroidism, osteitis fibrosa cystica, brown tumor, hypercalcemia, genu valgum

Introduction

Primary hyperparathyroidism (PHPT) is an endocrine disorder defined by the excessive release of parathyroid hormone (PTH), resulting in hypercalcemia (1). PHPT is rare in the pediatric population, with an estimated incidence of 2-5 cases per 100,000 children, compared to that in adults (2,3). PHPT is generally a sporadic condition, most commonly due to a single parathyroid adenoma or, less frequently, parathyroid hyperplasia (2). The incidence rates of PHPT are higher in women and in individuals of African origin compared to men and Caucasians, respectively (3). The clinical presentation may range from no symptoms at all to severe bone disease, including osteitis fibrosa cystica (OFC) (4). Patients with OFC can present with pathological fractures, bone pain and deformities, as well as radiological signs of severe hyperparathyroidism such as a salt-and-pepper skull, subperiosteal bone erosions, osteolytic lesions with sclerotic margins and profound demineralization (4). While PHPT may cause severe skeletal manifestations, surgical treatment, typically parathyroidectomy, can lead to significant improvement (5). However, alternative minimally invasive approaches, such as radiofrequency ablation, have emerged as potential treatment options, particularly in selected cases (6). Parathyroidectomy nonetheless remains the standard of care for symptomatic pediatric PHPT, and reported pediatric experience with radiofrequency ablation is very limited; the present case report should therefore be interpreted as an exceptional, carefully selected management approach rather than as support for its routine use.

Genu valgum (knock-knee deformity), as a presenting feature of pediatric PHPT with OFC is rare, with only a small number of documented cases (7). Although OFC was once prevalent in hyperparathyroidism, it currently only affects <2% of patients with PHPT (8,9). The present case report describes the case of a female child with PHPT with OFC who presented with genu valgum.

Case report

A 12-year-old girl presented to the Azadi Teaching Hospital (affiliated with the College of Medicine at University of Duhok),

Duhok, Iraq in November, 2023, with progressive genu valgum and experienced difficulty walking for 2 years. She reported generalized bone pain and constipation. An investigation of her previous medical history revealed an episode of acute pancreatitis associated with severe hypercalcemia (15.4 mg/dl; normal range, 8.5-10.6 mg/dl) requiring hospitalization. At that time, PTH was not measured, and she was discharged after recovery without further endocrine investigation. She subsequently underwent corrective osteotomy 12 months prior to the current presentation with no improvement in the genu valgum deformity. Upon an examination, she was found to have a short stature (height, 143 cm; 10th percentile) and bilateral genu valgum deformity without skeletal tenderness. Her family history was negative for hyperparathyroidism, nephrolithiasis, or other endocrinopathies, and a clinical evaluation did not reveal jaw tumors, pituitary lesions, or features suggestive of pancreatic endocrine neoplasia; however, formal genetic testing for MEN1, CDC73, RET, CASR and related genes was not performed, as this service was not available at Azadi Teaching Hospital. Her laboratory workup confirmed the diagnosis of PHPT (Table I).

An ultrasound of the neck revealed a well-defined vascular hypoechoic lesion of 13x26 mm in size located in the right infrathyroid area suggestive of a parathyroid adenoma (Fig. 1). X-rays of the lower limb revealed multiple cystic, expansile lesions with a 'soap-bubble' appearance in various areas (Fig. 2A and C).

The biopsy specimen from the left proximal tibial lesion was fixed in 10% neutral buffered formalin at room temperature (20-25˚C) for ~24 h, routinely processed and paraffin-embedded. Serial sections with a thickness of 4 μm were prepared using a rotary microtome and stained with hematoxylin and eosin (H&E; Bio-Optica) according to the manufacturer's protocol. A histopathological examination was performed using a bright-field light microscope (Olympus BX43, Olympus Corporation). The histopathological analysis revealed multinucleated giant cells, hemosiderin-laden macrophages, hemorrhagic areas and osteoid formation with osteoblastic rimming, findings consistent with a brown tumor secondary to hyperparathyroidism. The diagnosis was rendered by the institutional pathologist based on these morphological features interpreted in conjunction with the markedly abnormal biochemical profile; representative photomicrographs could not be reproduced, as archival histological slides were not retained at the Al-Mufty Medical-Laboratory. No additional immunohistochemical stains were performed.

Although surgical excision is the standard treatment for parathyroid adenoma, radiofrequency ablation was selected due to patient/family preference: Surgical parathyroidectomy was discussed with the family as the standard treatment, but the patient and her family declined surgery and specifically requested a minimally invasive alternative after counselling. The ablation procedure was performed by an experienced interventional radiologist (SHA) under ultrasound guidance; detailed intraoperative parameters (power, duration, and temperature monitoring range) were not systematically documented and are therefore not available for reporting. Close monitoring was performed for post-procedure hypocalcemia, and calcium supplementation was initiated to prevent hungry bone syndrome. Thereafter, the patient exhibited a marked improvement in her symptoms and the PTH level had decreased to 62 pg/ml (reference range, 15-65 pg/ml), while the serum calcium level decreased to 8.6 mg/dl (reference range, 8.5-10.6 mg/dl). Radiographs of the lower limbs revealed the near-complete resolution of the lesions (Fig. 2B and D) and the patient underwent removal of the plate and screw with her walking ability significantly getting better and marked clinical improvement of genu valgum on examination. A summary of the clinical timeline of the patient is presented in Table II.

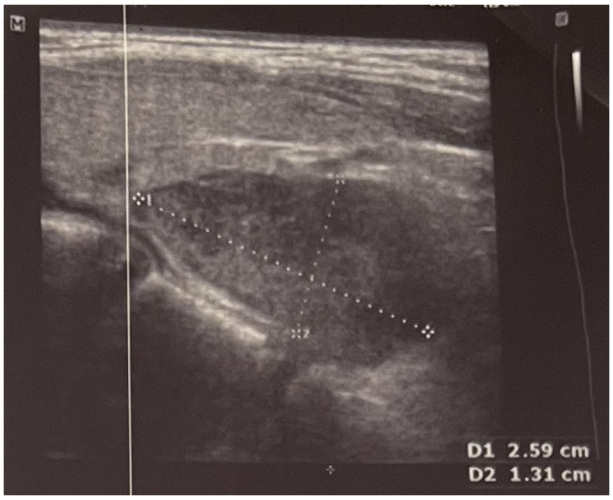

Figure 1. Ultrasound of the parathyroid glands demonstrating a well-defined vascular hypoechoic lesion of 13x26 mm in size located in the right infrathyroid area suggestive of a parathyroid adenoma.

Discussion

OFC is a severe skeletal manifestation of advanced PHPT (4). OFC was initially named as von Recklinghausen disease of the bone, highlighting the description by the German pathologist, von Recklinghausen in 1891 (10). The routine measurement of serum calcium levels in clinical practice with the resultant diagnosis of PHPT in patients with asymptomatic hypercalcemia, recognized since the 1970s, has led to the early treatment of such patients and a reduction in the incidence of OFC (6). The overall incidence of OFC has decreased from 69% to <2% of cases of PHPT (9). Although the case described herein suffered from a prior attack of acute pancreatitis likely due to severe hypercalcemia, she was not investigated for hyperparathyroidism, which led to a late diagnosis. This missed opportunity for diagnosis resulted in unnecessary orthopedic surgery and prolonged morbidity. It is critical to measure serum magnesium and creatinine levels in the setting of hyperparathyroidism, since patients with chronic hypomagnesemia or chronic kidney disease can develop secondary hyperparathyroidism with resultant bone resorption (11). The patient in the present case report had normal serum levels for both magnesium and creatinine. Radiological findings can be difficult to interpret and may mimic primary bone tumors or metastasis from other organs (5,7); thus, bone biopsy is typically required to

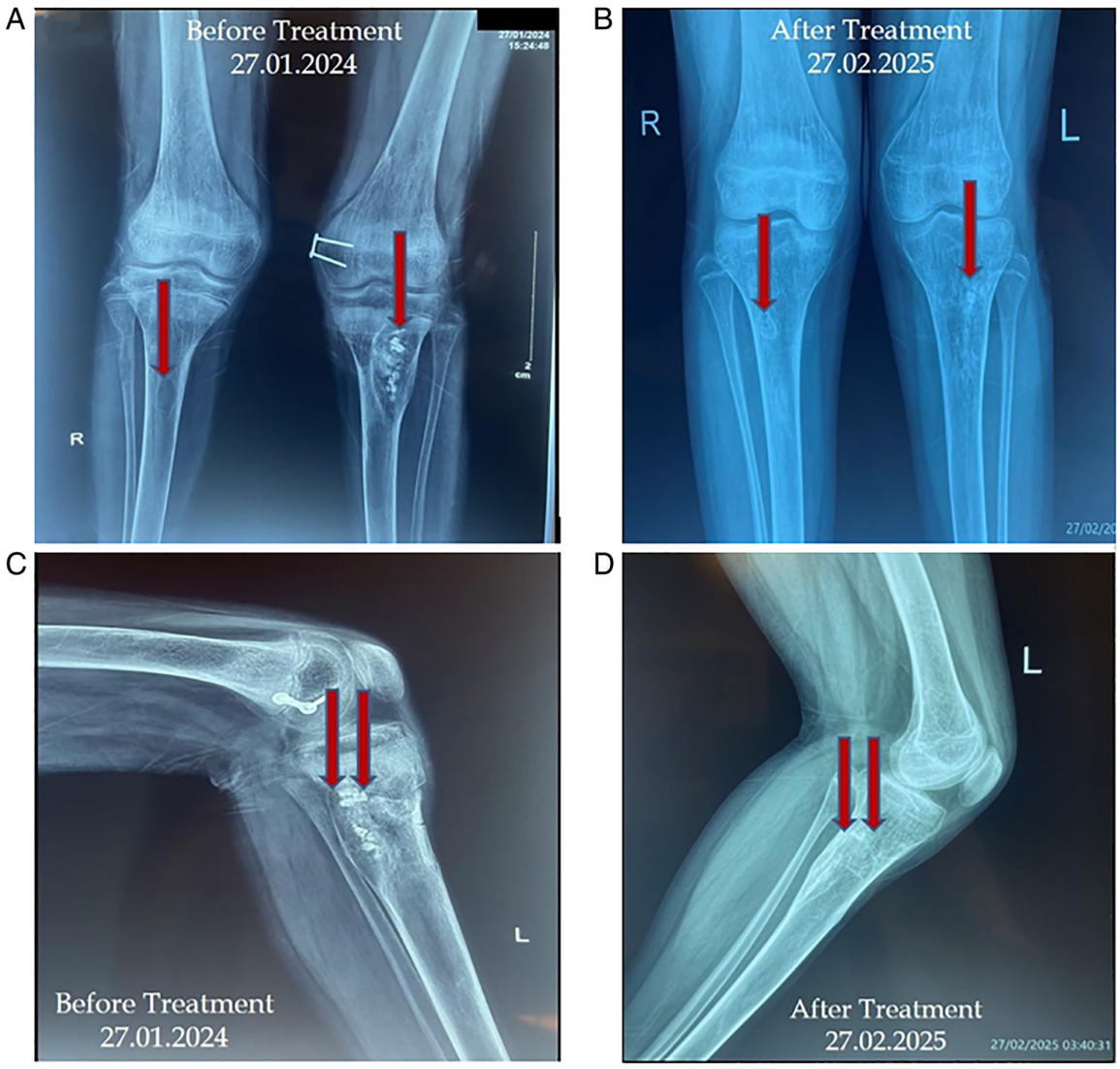

Figure 2. X-ray images of the lower limbs. (A and B) Anteroposterior view and (C and D) lateral view images. (A) Anteroposterior view illustrating a well-defined, expansile lesion in the proximal part of left tibia. The lesion exhibited a 'soap-bubble' appearance with cortical thinning and disruption of continuity, but no periosteal reaction. Another well-defined cystic lesion is shown in the proximal right tibia (red arrows). Overall bone density appears diminished. There is a plate and screw in the medial side of distal left femur. (C) Lateral view illustrating the lesion in the proximal area of the left tibia. (B) Anteroposterior view and (D) lateral view images following treatment demonstrating the almost-complete resolution of the lesions with significant mineralization compared to before treatment.

Table I. Results of the laboratory workup of the patient.

Parameter	Result	Reference range	Interpretation
Serum calcium	12.2 mg/dl	8.5-10.6 mg/dl	High
PTH	1,574 pg/ml	15-65 pg/ml	Very high
ALP	2,616 U/l	35-104 U/l	Markedly high
Vitamin D3	13.3 ng/ml	30-100 ng/ml	Low
Creatinine	0.2 mg/dl	0.3-0.7 mg/dl	Low-normal
Magnesium	2.3 mg/dl	1.5-2.5 mg/dl	Normal

PTH, parathyroid hormone; ALP, alkaline phosphatase.

confirm the diagnosis. While the gold-standard treatment of OFC is the surgical treatment of parathyroid disease (parathyroidectomy) (8,12), minimally invasive techniques have emerged as alternatives. Radiofrequency ablation has been

Table II. Clinical timeline of the patient.

Time point	Event
Baseline	Acute pancreatitis with severe hypercalcemia (15.4 mg/dl); PTH not measured; discharged without endocrine work-up
12 months prior to presentation	Corrective osteotomy for genu valgum; no improvement in deformity
Current presentation	Re-evaluation for persistent genu valgum and bone pain; short stature noted
Work-up	Laboratory confirmation of PHPT; neck ultrasound identifying parathyroid adenoma; lower-limb radiographs; tibial bone biopsy confirming brown tumor
Treatment	Radiofrequency ablation of parathyroid adenoma (surgery declined by family); hungry bone syndrome prophylaxis initiated
Follow-up	Biochemical normalisation of calcium and PTH; radiological follow-up showing near-complete resolution of lesions with remineralisation; hardware removal

PTH, parathyroid hormone; PHPT, primary hyperparathyroidism.

successfully used in adult PHPT (6). While genu valgum as a complication of OFC has been demonstrated (7,13,14), to the best of our knowledge, the literature regarding the use of radiofrequency ablation for pediatric PHPT is notably limited (6). The present case report adds to this literature, demonstrating successful ablation with the normalization of biochemical parameters and complete skeletal recovery. Parathyroidectomy nonetheless remains the gold-standard treatment for symptomatic pediatric PHPT. Given the very limited pediatric evidence base for RFA, the outcome of the case presented herein should be interpreted as an exceptional, patient-preference-driven management decision made in the absence of surgical consent, rather than as evidence supporting the routine use of radiofrequency ablation in pediatric PHPT. Prospective, systematically documented pediatric data, including standardized procedural parameters and orthopedic outcome measures, are warranted before the broader adoption of this approach may be considered.

In children and adolescents presenting with PHPT, a hereditary cause should always be actively considered. Family history and clinical features suggestive of MEN1-related manifestations (jaw tumors, renal lesions, pituitary or pancreatic endocrine tumors) were absent in the patient described herein; however, formal genetic testing for MEN1, CDC73, RET, CASR and related genes was not performed, as this service was not available at Azadi Teaching Hospital. It is recommended that genetic evaluation be strongly considered in all pediatric PHPT cases, regardless of an apparently sporadic presentation, given its implications for diagnosis, surveillance and family counselling (15).

The marked skeletal deformities in the case reported herein were treated with orthopedic surgeries rather than searching and treating the underlying cause. This highlights the critical importance of endocrine evaluation in children presenting with skeletal deformities, particularly when accompanied by other suggestive features, such as bone pain, growth failure, or prior hypercalcemia. Radiofrequency ablation was performed based on patient preference and resulted in a marked improvement in bone pain, mobility and the correction of genu valgum. The normalization of calcium and PTH levels further supports the success of the treatment. Of note, the reported case had vitamin D deficiency which is commonly observed in patients with PHPT. Vitamin D supplementation can be safely administered and will likely reduce the PTH level; however, the plasma calcium level should be monitored (16).

Several limitations of the present case report should be acknowledged: Representative histopathological images were unavailable; intraoperative ablation parameters and hungry bone syndrome prophylaxis dosing were not systematically documented; genetic testing for hereditary PHPT syndromes was not performed; sestamibi scintigraphy, 4D-CT and PTH washout were not available for localization confirmation; a subset of biochemical and imaging investigations (phosphate, corrected/ionized calcium, urinary calcium, renal ultrasound) were not obtained; and orthopedic outcomes were assessed clinically rather than with objective quantitative measures. These gaps limit the reproducibility and generalizability of the findings and are highlighted as priorities for prospective documentation in future pediatric PHPT cases managed with ablation.

In conclusion, the present case report highlights a rare pediatric presentation of PHPT with OFC and genu valgum, initially misdiagnosed as an isolated orthopedic issue. Early endocrine evaluation may have prevented unnecessary surgery and prolonged morbidity. Recognition of metabolic bone disease in children with unexplained skeletal deformities is crucial. Successful management of the underlying parathyroid pathology led to substantial improvement of skeletal abnormalities, although this should be regarded as an exceptional, carefully selected outcome rather than evidence supporting radiofrequency ablation as a routine alternative to parathyroidectomy in pediatric PHPT, pending further prospective data, emphasizing the need for interdisciplinary awareness among pediatricians, endocrinologists, and orthopedic surgeons.

Acknowledgements

Not applicable.

Funding

No funding was received.

Availability of data and materials

The data generated in the present study may be requested from the corresponding author.

Authors' contributions

All authors (BAQ, LOM, AMTA, SHA, HJA and AMJ) contributed to the conception and design of the study. Material preparation, data collection and analysis were performed by BAQ, SHA and HJA. BAQ was responsible for the diagnosis and advised on the treatment. SHA obtained ultrasound images and performed the radiofrequency ablation of the parathyroid adenoma. The original draft of the manuscript was written by HJA and AMJ. Revision and editing of the draft were performed by LOM and AMTA. BAQ and HJA confirm the authenticity of all the raw data. All authors have read and approved the final manuscript and agree to be accountable for all aspects of the work if questions arise related to its accuracy or integrity.

Ethics approval and consent to participate

The present case report was carried out in accordance with the Declaration of Helsinki. The parent/legal guardian of the patient provided written consent for the description of the present case.

Patient consent for publication

The parent/legal guardian of the patient described in the present case report provided written consent for the publication of the present case report and related images.

Competing interests

The authors declare that they have no competing interests.

References

1. Walker MD and Silverberg SJ: Primary hyperparathyroidism. Nat Rev Endocrinol 14: 115-125, 2018.
2. Marcocci C and Cetani F: Clinical practice. Primary hyperparathyroidism. N Engl J Med 365: 2389-2397, 2011.
3. Yeh MW, Ituarte PH, Zhou HC, Nishimoto S, Liu IL, Harari A, Haigh PI and Adams AL: Incidence and prevalence of primary hyperparathyroidism in a racially mixed population. J Clin Endocrinol Metab 98: 1122-1129, 2013.
4. Bandeira F, Griz L, Caldas G, Bandeira C and Freese E: From mild to severe primary hyperparathyroidism: The Brazilian experience. Arq Bras Endocrinol Metabol 50: 657-663, 2006.
5. Panagopoulos A, Tatani I, Kourea HP, Kokkalis ZT, Panagopoulos K and Megas P: Osteolytic lesions (brown tumors) of primary hyperparathyroidism misdiagnosed as multifocal giant cell tumor of the distal ulna and radius: A case report. J Med Case Rep 12: 176, 2018.
6. Issa PP, Kandil E and Lee GS: Radiofrequency ablation as a treatment modality for primary hyperparathyroidism: A systematic literature review. Gland Surg 13: 87-99, 2024.
7. Sakale H, Garg AK, Bhardwaj S and Agrawal AC: Genu valgum in children with primary hyperparathyroidism: A case series with a review of the literature. J Family Med Prim Care 13: 5912-5916, 2024.
8. De Crea C, Traini E, Oragano L, Bellantone C, Raffaelli M and Lombardi CP: Are brown tumours a forgotten disease in developed countries? Acta Otorhinolaryngol Ital 32: 410-415, 2012.
9. Vanitcharoenkul E, Singsampun N, Unnanuntana A and Sirinvaravong S: Osteitis Fibrosa Cystica and pathological fractures-the classic but neglected skeletal manifestation of primary hyperparathyroidism: A case report. BMC Musculoskeletal Disorders 22: 443, 2021.
10. Qasim BA: Historical landmarks in endocrinology and diabetes: The fascinating world of hormones. Newcastle upon Tyne: Cambridge Scholars Publishing, 2024.
11. Hamrah Siyani M, Zandiyeh F, Zaremoghadam E, Dehghan S, Sadeghian R, Aboulfathiyarmohammadyar Z and Alipoor M: Association of parathyroid hormone with plasma magnesium in health and disease; a review. J Parathyr Dis 11: e11211, 2023.
12. Bilezikian JP, Khan AA, Clarke BL, Mannstadt M, Potts JT and Brandi ML: The fifth international workshop on the evaluation and management of primary hyperparathyroidism. J Bone Miner Res 37: 2290-2292, 2020.
13. de Silva NL, Jayalath MD, Sampath WKC, Perera R and Karunathilake C: Primary hyperparathyroidism in an adolescent presenting with genu valgus progressing to extensive bone disease; a case report. BMC Endocr Disord 23: 71, 2023.
14. Boro H, Khatiwada S, Alam S, Kubihal S, Dogra V, Malla S and Kumar C: The spectrum of manifestations of primary hyperparathyroidism in children and adolescents. Pediatr Endocrinol Diabetes Metab 28: 178-187, 2022.
15. Starker LF, Akerström T, Long WD, Delgado-Verdugo A, Donovan P, Udelsman R, Lifton RP and Carling T: Frequent germ-line mutations of the MEN1, CASR, and HRPT2/CDC73 genes in young patients with clinically non-familial primary hyperparathyroidism. Horm Cancer 3: 44-51, 2011.
16. Yedla N, Kim H, Sharma A and Wang X: Vitamin D deficiency and the presentation of primary hyperparathyroidism: A mini review. Int J Endocrinol 2023: 1169249, 2023.