Journal of the ASEAN Federation of Endocrine Societies

CASE REPORT

Windswept Deformity: A Rare Skeletal Manifestation in an Adolescent with Primary Hyperparathyroidism

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Abstract

Primary hyperparathyroidism (PHPT) in adolescents is rare and has severe manifestations as compared to adults. Skeletal involvement in primary hyperparathyroidism in the form of deformities like genu valgus, genu varus and cubitus varus is rare and limited to case reports and case series. There is only one case of genu varus with genu valgus on the contralateral extremity (windswept deformity) that has been reported to date in the literature. We report the case of a 19-year-old male who presented with isolated progressive bending of his legs at the knee (windswept deformity) for three years. He was found to have hypercalcemia, hypophosphatemia, high alkaline phosphatase, high intact parathyroid hormone (iPTH), normal 25-hydroxy vitamin D level and a normal kidney function test. A diagnosis of primary hyperparathyroidism was made. On imaging studies, a left inferior parathyroid adenoma was localized and was successfully removed surgically. Serum calcium and iPTH normalized post-operatively. The patient is being planned for corrective osteotomy after stabilization of alkaline phosphatase levels.

Key words: primary hyperparathyroidism, skeletal manifestations, genu valgus, genu varus, wind-swept deformity

INTRODUCTION

Primary hyperparathyroidism (PHPT) is rare in pediatric and adolescent-aged individuals with an incidence of 1/300,000 live births/ year and a prevalence of 3-5/100,000.[1] Single gland disease is the most common presentation that is similar to adults. It can also be a part of syndromes like multiple endocrine neoplasia (MEN).[2] In developed countries, it is diagnosed earlier because of more frequent serum calcium screening.[3] However, florid manifestations, including skeletal manifestations with deformities, are still seen in developing countries like India.[4] Skeletal deformity, especially windswept deformity (genu valgus in one limb and genu varus in contralateral limb), in PHPT is rare. The common differentials for this deformity are rickets (hypophosphatemia, vitamin D deficiency and calcium deficiency), skeletal dysplasia, chronic fluoride toxicity, distal renal tubular acidosis, renal osteodystrophy and, rarely, trauma.[5] If PHPT and associated deformity are not diagnosed early and promptly treated, this can lead to further deformities, gait abnormalities, limb shortening and osteoarthritis.[6] We report a 19-year-old male who presented with windswept deformity due to primary hyperparathyroidism from a parathyroid adenoma. Early diagnosis of PHPT is important to prevent significant morbidity in the form of end-organ damage and mortality.

CASE

A 19-year-old male presented with progressive bending of his legs at the knee over three years' duration which his parents attributed to excess body weight. The deformity worsened in the last six months for which he was brought to the orthopedic outpatient department (OPD) of our institute. He had no history of pain, swelling, or trauma to the knees. On laboratory investigations, he was found to have hypercalcemia which prompted an endocrinology service referral. He had achieved developmental milestones at the appropriate age. His two younger brothers and one elder sister did not report any similar deformities. There was no significant personal or family history of renal calculi or multiple endocrine neoplasia (MEN)-related disorders. On examination, his height was 167 cm (10^{th}-25^{th} percentile), weight was 77 kg (75^{th}-97^{th} centile) with a computed body mass index of 27.6 kg/m^2 (obese). He was Tanner's stage 5 in pubertal development. A windswept deformity was noted in the lower limbs (genu valgus in the right lower limb, and genu varus in the contralateral lower limb) (Figure 1). Clinically, there were no other skeletal deformities that were noted. Laboratory evaluation revealed elevated albumin-corrected serum calcium, low serum phosphate, high alkaline phosphatase, high intact parathyroid hormone (iPTH), normal 25-hydroxyvitamin D levels and a normal

eISSN 2308-118x (Online)
Printed in the Philippines

Received: December 20, 2023. Accepted: February 27, 2024.
Published online first: August 1, 2024.
https://doi.org/10.15605/jafes.039.02.10

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Figure 1. Windswept deformity (genu valgus on the right lower limb and genu varus on the left lower limb) on clinical examination of the patient.

Table 1. Baseline laboratory profile at the time of endocrine evaluation

Parameters	Patient's pre-operative values	Reference range
Serum creatinine (mg/dL)	0.5	0.66 -1.25
Serum albumin (g/dL)	4.2	3.5-5
Serum albumin-adjusted total calcium (mg/dL)	11.9	8.4-10.2
Serum phosphate (mg/dL)	2.7	2.7-4.7
Serum alkaline phosphatase (U/L)	2929	38-126
Serum intact Parathyroid Hormone (iPTH) (pg/ml)	720	7.5-53.5
Serum 25-hydroxyvitamin D (ng/ml)	28.8	30-100
Serum magnesium (mg/dL)	2.4	1.6-2.5

kidney function test (Table 1). The results were consistent with a diagnosis of primary hyperparathyroidism. Ultrasonography of the neck showed a hypoechoic lesion in the inferior pole of the left thyroid lobe suggestive of a parathyroid adenoma. Technetium (^{99m}TC) tetrofosmin scan revealed a left inferior parathyroid adenoma (Figure 2). There was no evidence of gallbladder or renal calculi on ultrasonography. Genetic testing for MEN syndromes could not be done because of non-availability at our Institute. A skeletal survey revealed valgus deformity at the right knee joint and varus deformity at the left knee joint (Figure 3). There was generalized osteopenia with subperiosteal resorption of proximal phalynx of the first finger, middle phalanx of the second, third, fourth and fifth fingers of both hands and brown tumor in the middle phalynx of the fourth finger of the left hand and and acro-osteolysis in the distal phalanx of the second, third, and fourth finger of the left hand (Figure 4). Interestingly, radiographs of both hands revealed open physes of both the radius and ulna. Subsequently, the patient underwent a left inferior parathyroidectomy. Repeat serum intact PTH level 10 minutes after left inferior parathyroid gland excision decreased by >50% from 720 pg/mL to 46.2 pg/mL, suggestive of cure. Histopathology confirmed the excised tissue to be a parathyroid adenoma. Postoperatively, he developed tingling sensations in the perioral area with positive Chvostek's sign. Laboratory evaluation showed total calcium of 7.6 mg/dL (albumin-corrected calcium of 7.5 mg/dL) and serum phosphate of 2 mg/dL possibly because of post-parathyroidectomy hungry bone syndrome. Treatment with intravenous calcium gluconate and oral calcitriol was given for two days. From the third postoperative day onwards, treatment with oral calcium carbonate 2 grams per day and oral calcitriol 0.5 micrograms twice a day was continued. The serum intact PTH on day seven of surgery was 53 pg/mL and albumin-corrected total calcium was 9.6 mg/dL. Monthly cholecalciferol (60000 IU) and daily calcium carbonate supplementation were continued. Corrective osteotomy is being planned by the orthopedic team after stabilization of the serum alkaline phosphatase level. In the meantime, he has been advised lifestyle modification for weight reduction to decrease mechanical pressure on lower limbs to prevent worsening of deformity.

DISCUSSION

Primary hyperparathyroidism in children and adolescents is uncommon. In addition, because of nonspecific signs and symptoms of the disease, healthcare professionals may fail to check serum calcium levels which will further delay the diagnosis and predispose patients to present with end-organ pathology.[7] Renal stone disease, bone involvement, and nephrocalcinosis are among the most common end-organ pathologies in pediatric patients.[8] Skeletal diseases associated with PHPT include bone pain, osteopenia, fractures or lytic bone lesions and bone deformities.[9]

Skeletal deformities in the form of genu valgus, genu varus, genu valgus on one side and genu varus on the contralateral side (windswept deformity) and cubitus valgus from primary hyperparathyroidism in children and adolescents are uncommon. Though they have been reported in the literature, the presence of these deformities should be assessed with a high index of suspicion for metabolic causes and should necessitate further workup. The typical causes of windswept deformity are rickets (hypophosphatemic, vitamin D deficiency, calcium deficiency), skeletal dysplasias, chronic fluoride toxicity, distal renal tubular acidosis, renal osteodystrophy and, rarely, trauma.[5] Timely identification of its etiology and corresponding treatment is important. Treatment options, apart from treatment of the specific etiology, include surgical (corrective osteotomies, stapling) or conservative (plaster casting) approaches. If left untreated, it can lead

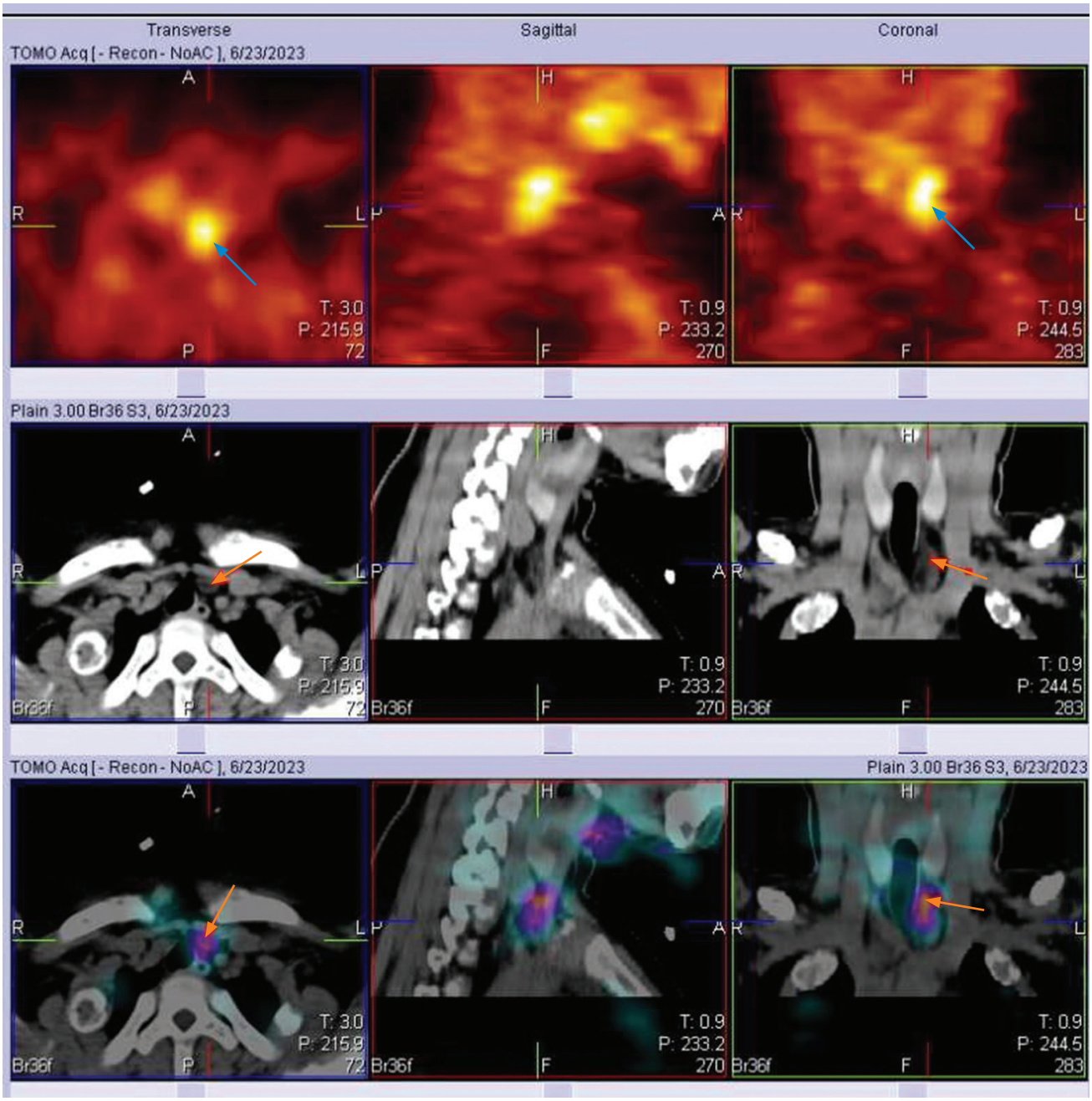

Figure 2. Technetium (^{99m}Tc) tetrofosmin and SPECT/CT scan showing tracer accumulation in the region of the left inferior parathyroid gland suggestive of left inferior parathyroid adenoma.

to further deformities, gait abnormalities, limb shortening and osteoarthritis.

To our knowledge, there are only 36 cases of genu valgus, one case of cubitus valgus, and one case each of genu varus and windswept deformity have been reported in the literature (Table 2). The age of presentation of cases presented ranged from 11 to 21 years. The pathogenesis of such deformities in this age group is unclear. However, it has been postulated that elevated parathyroid hormone can directly affect growth plate and bone remodeling during pubertal growth spurt.[10] We propose that the reported patient's excess body weight, in addition to the effect of PTH, may have contributed to the deformity.

In addition to genu valgus, other radiological features typical of primary hyperparathyroidism have also been reported in cases reported in the literature. These include osteopenia, subperiosteal resorption of phalanges, acro-osteolysis, brown tumor, and salt-and-pepper appearance of a skull.[9-33] 16 patients had features of rickets clinically or radiologically. Our patient presented with windswept deformity in the lower limb without other symptoms due to hypercalcemia and no characteristic features of rickets. He had radiological features of primary hyperparathyroidism similar to the cases reported in the literature. He had a single parathyroid adenoma similar to all reported cases in the literature (Table 2).

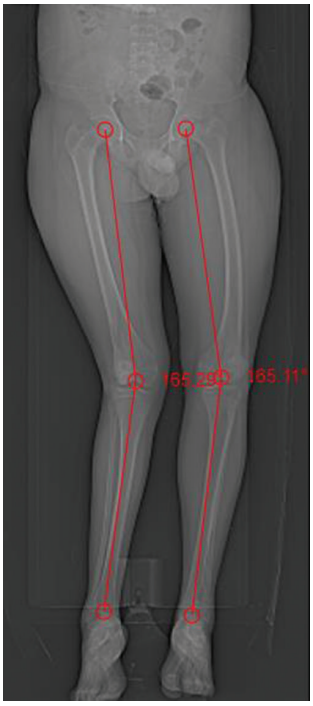

Figure 3. Radiograph of bilateral tibia and femora showing windswept deformity at the knee, characterized as genu varus on the left side (15°) and genu valgus (15°) on the right side.

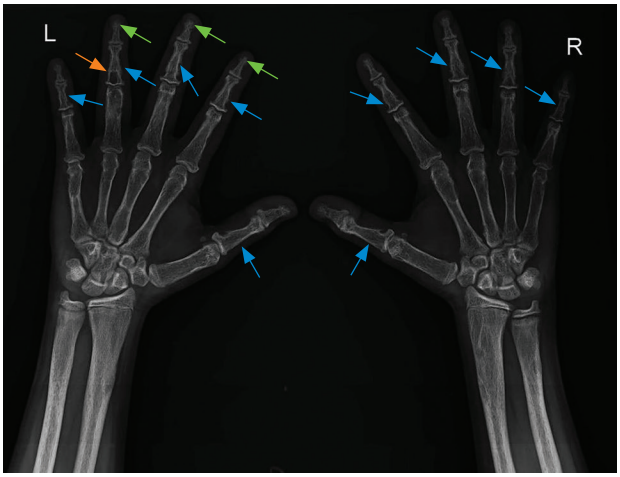

Figure 4. Radiograph of both hands showing generalized osteopenia with subperiosteal resorption *(blue arrows)* of the middle phalanx of the first, second, third, fourth and fifth fingers of both hands, brown tumor *(orange arrow)* in the middle phalanx of the fourth finger of the left hand and acro-osteolysis *(green arrows)* in the distal phalanx of the second, third and fourth finger of the left hand.

Table 2. Summary of reported cases with primary hyperparathyroidism and skeletal deformities

Author, Year	Age (years), Sex	Clinical feature	Etiology
De Silva et al., 2023[11]	18, M	Bilateral (B/L) genu valgus, short stature	LIPA
Dikova et al., 2021[12]	12, F	B/L genu valgus, inability to walk	RIPA
	15, F	Left genu valgus, waddling gait	LIPA
Boro H et al., 2022[9]	15, F	Bone pain, genu valgus	LIPA
	17, F	Fatigue, genu valgus, bone pain	LIPA
	19, F	Bone pain, right humerus fracture, reflux symptoms, genu valgus	RIPA
	15, M	Fatigue, genu valgus, renal stones	LIPA
	17, M	Bone pain, genu valgus, cubitus valgus, constipation	RIPA
Lee et al., 2021[13]	15, M	B/L genu valgus, short stature	LIPA
Yanrismet Y et al., 2019[14]	13, M	B/L genu valgus, bone pain, muscle weakness	RIPA
Rao KS et al., 2019[15]	12, F	B/L genu valgus, renal stones	RIPA
Paruk IM et al., 2019[16]	17, M	B/L genu valgus, short stature	LIPA
	13, M	Left genu valgus, right genu varus	RIPA
Khan et al., 2019[17]	17, M	B/L genu valgus, short stature, muscle wasting	LIPA
George GS et al., 2019[18]	15, M	B/L genu valgus, Slipped capital femoral epiphysis	RIPA
Kamath SP et al., 2018[19]	11, F	Fatigue headache, B/L genu valgum	LIPA
	12, M	B/L genu valgum, renal stones	LIPA
Pradhan R et al., 2018[20]	15, F	B/L genu valgum, widening of wrists	LIPA
	15, F	B/L genu valgum fractured left clavicle, kyphosis, rachitic rosary	-
	11, M	B/L genu valgum, proximal muscle weakness	-
Arambewela MH et al., 2017[21]	12, F	B/L genu valgus	RIPA
Zil-E-Ali A et al., 2016[22]	14, F	Short stature, B/L genu valgus, pectus carinatum, scoliosis	RIPA
Sharma S et al., 2016[23]	15, F	Genu valgus, pectus carinatum	LIPA
Ganie M et al., 2016[24]	14, M	B/L genu valgus, bone pain	LIPA
	14, M	Genu valgus, Widening of wrists	Right lower neck (ectopic) adenoma
	15, M	Genu valgus, bone pain, fracture of right tibia	RIPA
Ramkumar S et al., 2014[10]	16, M	Leg pain, B/L genu valgus, generalised arthralgia, polyuria	LIPA
	13, M	B/L genu varus, Nausea, abdominal pain	RIPA
Ratnasigam J et al., 2013[25]	15, F	B/L genu valgus	Right parathyroid adenoma
Dutta D et al., 2013[26]	12, F	B/L genu valgus, short stature, flat feet	RIPA
Walczyk A et al., 2011[27]	15, M	B/L genu valgus, seizures	RIPA
Harman CR et al., 1999[8]	14, F	B/L genu valgus	-
Menon PS et al., 1994[28]	14, F	B/L genu valgus, short stature, renal stones, rachitic rosary	LSPA
Kauffmann C et al., 1993[29]	13, F	B/L genu valgus	LIPA
Rapaport D et al., 1986[30]	15, F	B/L genu valgus, renal stones	RIPA
	15, M	B/L genu valgus, renal stones	RIPA
Lloyd HM et al., 1965[31]	14, M	B/L genu valgus, weakness, scoliosis	LIPA
Balch HE et al., 1953[32]	21, F	B/L genu valgus, vomiting, clubbing, rib tenderness	LIPA
Mcclure RD et al., 1945[33]	14, F	B/L genu valgus	LIPA

LIPA – Left inferior parathyroid adenoma, RIPA – Right inferior parathyroid adenoma, LSPA – Left superior parathyroid adenoma, M – Male, F – Female, B/L – Bilateral

Surgical removal of the involved parathyroid gland is the mainstay of treatment of PHPT.[7] Our patient underwent a successful parathyroidectomy, similar to the reported cases, and he is currently being planned for osteotomy for windswept deformity correction.

CONCLUSION

We have reported a rare case of skeletal manifestation in the form of genu varus in one limb with genu valgus in the contralateral limb (windswept deformity) in a male adolescent secondary to PHPT. As PHPT is rare in adolescents and may mimic other conditions, like rickets, when the disease manifests in the form of skeletal deformities, misdiagnosis and inappropriate management may inevitably lead to further aggravation of skeletal system involvement. When children and adolescent patients present with skeletal manifestations and deformities, this should prompt the immediate diagnostic evaluation by measuring serum calcium, phosphorus and PTH levels to make a timely diagnosis and treatment. Prompt treatment may eventually lead to a cure and prevent end-organ damage. Moreover, additional research is needed to further understand the pathogenesis of skeletal deformities in PHPT in children and adolescent patients.

Caregiver's Perspective
Before coming to JNMC and DMIHER at Sawangi, my wife and I were overwhelmed. Our son's diagnosis left us confused, afraid and emotionally drained. The financial burden added a heavy weight to our worries. Thankfully, the doctors here at JNMC and DMIHER provided us with the cooperation, proper evaluation, and effective management we desperately needed. We are happy to say that our son's treatment is progressing well.

Ethical Consideration
Patient consent was obtained before submission of the manuscript.

Statement of Authorship
All authors certified fulfillment of ICMJE authorship criteria.

CRediT Author Statement
PF: Conceptualization, Writing – original draft preparation; **BJ:** Writing – review and editing; **SS:** Writing – review and editing, Supervision.

Author Disclosure
The authors declared no conflict of interest.

Data Availability Statement
No datasets were generated or analyzed for this study.

Funding Source
None.

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