

Presentation of parathyroid adenoma with genu valgum and thoracic deformities

Ahsan Zil-e-Ali,¹ Aiza Latif,² Anam Rashid,³ Asim Malik,⁴ Haseeb Ahmed Khan⁵

Abstract

Parathyroid adenoma is the main cause of primary hyperparathyroidism. It is usually asymptomatic and occurs more commonly in adults. It presents with raised parathormone (PTH) and Ca⁺ levels in serum. Its presentation in adolescence is rare. We report one such incidence of a 14 years old girl who presented with bone pains short stature, and generalized muscle wasting. She was found to have genu valgum at the knee joint, pectus carniatum, scoliosis and cystic changes in pelvis and calvarium. Biochemical investigations and parathyroid Tc-99mMIBI scan confirmed the diagnosis of a parathyroid adenoma. The gland was removed by parathyroidectomy. Till date 12 such cases are reported and none had thoracic, vertebral or calvarium involvement.

Keywords: Endocrinology, Pediatrics, Orthopedics, Parathyroid Adenoma, Radiology.

Introduction

The major function of parathyroid gland is to secrete parathormone (PTH), a key regulator of serum calcium levels and concurrently affecting the bone calcium. Physiologically, PTH acts on distal convoluted tubule of nephron to increase calcium retention. Clinically hyperparathyroidism presents with increased PTH and hypercalcaemia, mostly it is asymptomatic and hard to diagnose. A specialized approach is needed to diagnose and to manage such cases.¹ Parathyroid adenoma is a rare case in younger age groups and often a solitary adenoma is developed, although spectrum of presentations has been reported. These signs and symptoms may vary according to age and race.² Parathyroid adenoma presents with primary hyperparathyroidism which has characteristic findings of bone fractures and urinary calculi.³ Overall, primary hyperparathyroidism is not a common disorder in adolescents and so far 12 such cases are reported. None of these cases was reported with changes in thoracic skeleton, scoliosis, pelvis or calvarium.

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¹⁻³FMH College of Medicine, ⁴Department of General Surgery & Minimally Invasive Surgery, ⁵Department of Pathology, Fatima Memorial Hospital, Shadman, Lahore.

Correspondence: Ahsan Zil-E-Ali. Email: ahsanzileali@gmail.com

These days the diagnosis and prompt management is available, reducing further complications.⁴

Here a case of a 14 years old girl is reported, presenting with genu valgum and thoracic deformities. She was diagnosed to have raised intact parathormone (iPTH), high serum calcium levels and pectus carniatum due to right lower parathyroid adenoma which was resolved after parathyroidectomy.

Case Report

In March 2014, a 14 years old girl presented with a normal perinatal history and developmental milestones, genu valgum (Figure-1), severe pain in knee joints which was progressive in nature and difficulty in walking. The symptoms of pain appeared at the age of 12 years along with easy fatigability, malaise and bowing of legs.

The patient had short stature with weight of 35 kg, and was underweight with BMI of 18.12 [height: 139 cm, Standard deviation Score: -0.5, target height: 168cm, Z-score: -56.60]. Her menarche was at 12 years of age. On general physical examination, it was observed that patient was tachycardic (101 beats/minute) although no murmur was heard on auscultation. Genu valgum and absence of foot arches with

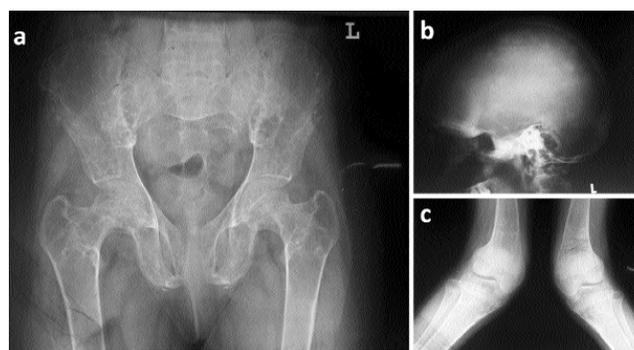


Figure-1: (a) Radiograph of pelvis with frontal projection showing generalized decrease in bone density. Lytic or sclerotic lesion is noted. Both hip joints are normal however sacroiliac joints appear to be ankylosed. (b) A radiograph of a skull showing cystic formations with lytic and sclerotic sites on the table. No reactive sites are seen. (c) Both knee frontal projection showing reduction of medial joint space bilaterally and symmetrical along with angulation of femur, tibia and fibula resulting in valgus deformity. No periosteal reaction, no lytic or sclerotic lesion seen. Soft tissues are unremarkable.

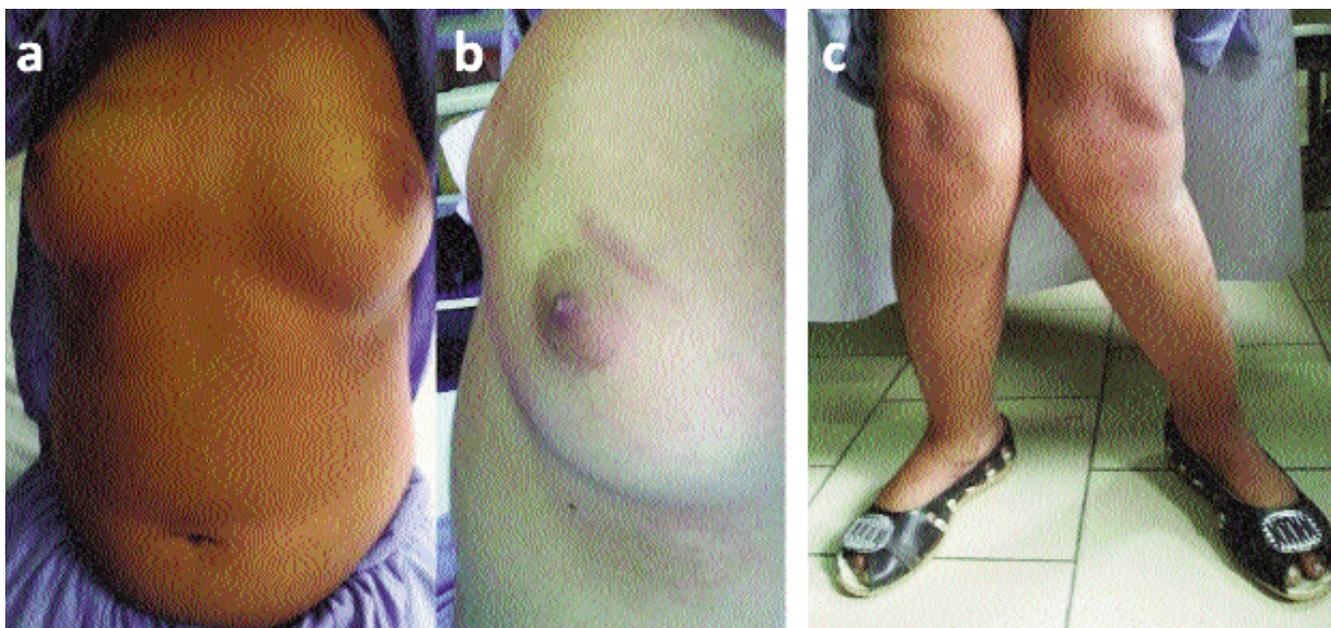


Figure-2: Image showing chest deformation, showing pectus carinatum, Front View (a), Lateral View (b), Genu Valgum with splaying below the knee joint (c).

Table-1: Major laboratory investigations, pre and post-surgical.

Parameters	Pre-surgical report	Postsurgical 8th day report
Serum Calcium	12 mg/dl	7.6 mg/dl
Phosphorus	2.0 mg/dl	3.6 mg/dl
iPTH	3203 pg/dl	53pgdl
Alkaline Phosphatase	415 IU/L	92 IU/L
SGPT	42 IU/L	38 IU/L
ESR	55 after 1 hr	22 after 1 hr
25-hydroxyvitaminD	24.9 ng/ml	28.7 ng/ml

iPTH: Intact parathormone.

SGPT: Serum Glutamic Pyruvic Transaminase.

ESR: Erythrocyte Sedimentation Rate.

generalized muscle wasting were also noted. On further evaluation of thorax, it was seen that there was twisting of the backbone from the midline forming scoliosis and there was a markedly raised sternum forming a classical pectus carinatum (pigeon chest) (Figure-2).

Skeletal radiography revealed splaying of lower limb below the knee joint, decreased bone mineral density, thin cortices and several cystic brown tumours in pelvis and calvarium presenting the initiation of von Recklinghausen disease of bone (Figure-1).

On a full length computed radiograph, it was evident that the patient had a narrower angle of inclination at neck of femur, right 110.3o, left 112.4o and shorter length of

lower limbs, 738mm. Biochemical investigation revealed mild deficiency of cholecalciferol, there was no dietary supplementation in the past. On electrolyte evaluation, hypercalcaemia was revealed with a raised value of 13.5 mg/dl (Normal: 9-10.5mg/dl). Intact Parathormone (iPTH) report was suggestive of hyperparathyroidism with the value of 3203pg/ml (Normal: 10-55 pg/ml). There was also a slight increased Tri-iodo-thyroidine level (T3) although its relevance was not made with hyperparathyroidism.

All of the liver function tests were normal but alkaline phosphatase was highly raised, which was evidence for hyperparathyroidism with such presentations. Complete blood count was normal and ESR was 55 at the end of 1st hour. Renal function tests were normal.

Urine analysis revealed that urine was very dark coloured with routine traces of epithelial and pus cells. On further investigation the ^{99m}Tc MIBI (methoxyisobutylisonitrile) dual phase parathyroid scan was done using 750 MBq and images were acquired at a durations of 30 minutes, 1 hour and 2 hours. An abnormal increased uptake was persistent on delayed images at lower pole of thyroid lobe which was confirmatory for right lower parathyroid adenoma.

The patient underwent right lower parathyroidectomy and its histopathology report showed abundant clear cells and chief cells in a disarray with increased nucleus to cytoplasmic ratio, neutrophilic infiltrate and a rim of compressed normal parathyroid

parenchyma with intact capsule.

After parathyroidectomy, the value of serum calcium level markedly reduced on the 8th day. For correction of genu valgum, a surgical procedure of medial close wedged osteotomy at distal end of femur was done. It was planned by the surgical team that scoliosis should not be operated; as correction of it may lead to extensive paralysis. Parathyroidectomy was promising as the serum iPTH dropped to normal, indicating that no further deformity would occur.

Discussion

Parathyroid adenoma is the major cause of primary hyperparathyroidism. It is a rare disease during adolescence however it is common in adults. It is asymptomatic on general routine examinations although on biochemical investigations it can be suspected after raised serum calcium levels (>5.6 mg/dL) and raised iPTH. The underlying physiology of parathormone exacerbates into a disease which affects various organic systems in the body. Involvement of the bone with raised parathormone isn't rare but our case had a very peculiar presentation with involvement of the knee joint, vertebrae, pelvis and thoracic skeleton. The patient was underweight with a low body mass index as mentioned before. It can be assumed that genu valgum can be a consequence of weight bearing by weakened bones.

Twelve cases have been reported before and none presented with thoracic skeleton deformities or cystic changes in the calvarium. All the cases reported so far had genu valgum, solitary parathyroid adenomas and none of the cases have an association with multiple endocrine neoplasia (MEN-1/MEN-2A). All cases ever reported are in range of 13 to 16 years, only one case was of 21 years old. Ten cases had normal vitamin D and three out of all these cases also had gastrointestinal symptoms. Only two cases were reported with body mass index that were in normal range.⁵

A similar case of a 27 year old lady was presented by Amin et al, with swelling on mandible and molar involvement. Biopsy showed that it was a giant cell granuloma and electrolyte investigation reports showing raised serum calcium. By the reports, the suspicion of malignancy was ruled out though on further evaluation, PTH was raised but unexpectedly levels of alkaline phosphatase was in range.⁶

Another rare presentation was reported by Tahim et al.⁷ A 53 year old man presented with gastroesophageal reflux

symptoms, epigastric pain and vomiting. On cardiovascular examination, the patient was in tachycardia with hypotension, though he had a normal sinus rhythm on EKG. On laboratory reports, alkaline phosphatase was raised and had normal Vitamin D. There was no bone related pathology.⁷

According to Cakal et al,⁸ high resolution ultrasonography (USG), Tc 99m- Methoxyisobutylisonitrile parathyroid scintigraphy and magnetic resonance imaging (MRI) are the leading options for radiologic diagnosis. MRI is reported to have the highest positive predictive value. These tumours are surgically treated under local anaesthesia. Minimal tumour invasion and unilaterally denote a good surgical outcome.⁸

Conclusion

Our case has added a new query to the subject of endocrinology and further research is needed to find peculiarities and presentation in a parathyroid adenoma. If this tumour is detected early and removed, it is possible that such skeletal deformities can be minimized. A clinician in a primary healthcare setup should also keep parathyroid adenoma in differential diagnosis of skeletal abnormalities, till it is ruled out. Further evidence based knowledge is needed in this perspective.

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