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Dr. Sushant Nadkarni
Post Graduate, Department of Orthopaedics, KIMS Hubli, Karnataka, India

Dr. Suryakanth K
Professor and Unit Head, Department of Orthopaedics, KIMS Hubli, Karnataka, India

Dr. AA Hosangadi
Associate Professor, Department of Orthopaedics, KIMS Hubli, Karnataka, India

Corresponding Author:
Dr. Sushant Nadkarni
Post Graduate, Department of Orthopaedics, KIMS Hubli, Karnataka, India

Primary hyperparathyroidism in an adolescent girl masquerading as rickets: A diagnostic challenge

Dr. Sushant Nadkarni, Dr. Suryakanth K and Dr. AA Hosangadi

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Abstract
Introduction: Primary heperparathyroidism usually presents at 5th-6th decade of life with its classical triad of 'stones, bones and groans. It is uncommon in children and adolescents which when presented at this age can mimic rickets and leads to misdiagnosis.
Materials and Methods: Case was of 14 year old girl who presented with bilateral progressive genu valgus deformity with proportionate short stature and difficulty in walking. Preliminary x-rays of both knee joints showed tibio-femoral angles of 19° right and 17° left respectively and x-rays of bilateral wrists and ankle showed features consistent with rickets. Biochemical evaluation showed normal serum calcium levels initially which was elevated after vitamin D supplementation that was given for sub-optimal vitamin D levels. Serum parathormone levels were significantly elevated at presentation and even after vitamin D and calcium supplementation. Urine calcium and phosphate levels were within normal range. Four-dimensional [4D] CT of neck revealed hypodense, heterogenously enhancing well defined lesion posterosuperior to left thyroid gland suggesting parathyroid adenoma. Ultrasound guided aspiration was done which confirmed parathyroid adenoma histologically. Patient underwent left superior parathyroidectomy.
Conclusion: Evaluation of rickets by clinical, radiological, biochemical and histological parameters led to the diagnosis of primary hyperparathyroidism which was due to parathyroid adenoma. Hence, skeletal deformity resembling rickets in adolescents entails detailed evaluation to unmask parathyroid adenoma for the appropriate diagnosis and treatment.

Keywords: Primary hyperparathyroidism, parathyroid adenoma, rickets

Introduction
Primary heperparathyroidism is the leading cause of metabolic bone disease and hypercalcaemia at 5th-6th decade of life with its classical triad of 'stones, bones and groans and it is rare in children and adolescents with the estimated incidence of 2-5/100,000 live birth [1]. The most common cause of primary hyperparathyroidism are adenomas and hyperplasia and rarely carcinomas of the parathyroid gland. Primary hyperparathyroidism when presented in children and adolescents can mimic rickets in terms of clinical and radiological parameters and can lead to misdiagnosis. We report a case of an adolescent girl who presented with typical features of rickets but on subsequent evaluation, diagnosed as parathyroid adenoma which was masked by her skeletal and radiological manifestations.

Materials and Methods
A 14 year old girl with normal perinatal and developmental history presented with bilateral progressive genu valgus deformity, difficulty in walking with proportionate short stature and lack of development of secondary sexual characters. The leg deformity was started at 12 years of age which limited her outdoor activities. The patient was significantly undernourished body weight-22.7kgs which was below 3rd centile. Short stature was evaluated and it showed height 130cm upper segment-63cm, lower segment-67cm which was below 3rd centile according to WHO growth chart.

On head to toe examination, there was frontal bossing, pegion chest, rhichitic rosary, widening of bilateral wrists Figure 1, knocking of knees, visible thigh and calf muscle wasting bilaterally Figure 2 and proximal lower limb muscle weakness which made the patient walk with hand to knee gait. Patient was prepubertal according to Tanner sexual maturity staging. Radiographic evaluation showed widening of distal end of radius, ulna with osteopenia Figure 3.

Scannogram of lower limb showed tibio-femoral angles of 19° right and 17° left, Figure 4 respectively. Initial biochemical values were suggestive of vitamin D deficiency rickets with sub-optimal levels of vitamin D, normal serum calcium levels and elevated levels of parathormone and alkaline phosphatase which on subsequent treatment with vitamin D 60,000 U weekly along with calcium supplements resulted in normal values of 25-hydroxycholecalciferol, elevated serum calcium levels and further elevation of parathormone levels. This led to the suspicion of alternative cause for hyperparathyroidism and four-dimensional 4D CT of neck was done which showed hypodense, heterogenously enhancing well defined lesion measuring 2.1×1.5cm noted on left side of neck, posterior to posterolateral surface of left thyroid gland Figure 5 in tracheoesophageal groove Figure 6 suggestive of parathyroid adenoma. Fine needle aspiration cytology of the left superior parathyroid gland was done which showed round to oval cells with clear cytoplasm arranged in clusters Figure 7, loosely cohesive sheets in microfollicular pattern with no evidence of atypia and malignancy suggestive of chief cell predominant parathyroid adenoma. Patient underwent left superior parathyroidectomy and a 3×2 cm smooth, encapsulated, ovoid shaped parathyroid adenoma was resected Figure 8. Serial calcium level monitoring was done in the post operative period and calcium gluconate was given for hypocalcaemia, which attained normal levels after one week. Post operative serum parathormone level was significantly low Table 1. Following parathyroidectomy, bone and joint pain were reduced, proximal muscle weakness was resolved and the quality of life improved.

Fig 1: Showing widening of bilateral wrist joints

Fig 2: Showing knocking of knees, wasting of calf muscles and widening of ankle joints

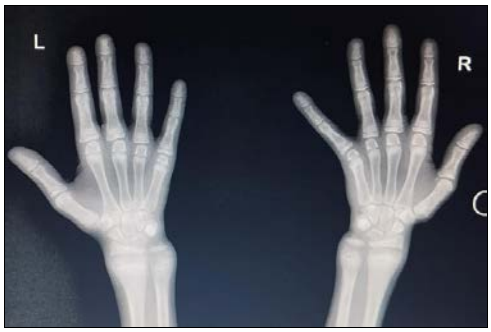

Fig 3: Radiograph of bilateral wrist showing osteopenia with widening of distal end of radius and ulna at the metaphysis

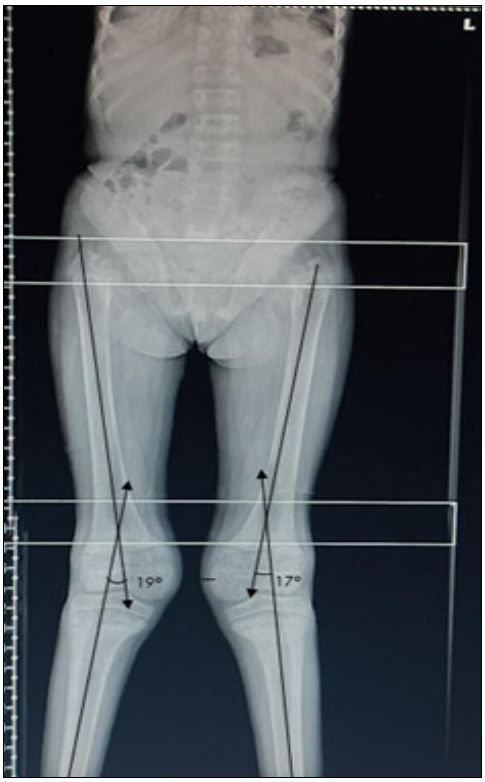

Fig 4: Scannogram of bilateral lower limbs showing tibio-femoral angles of 19° right and 17° left respectively

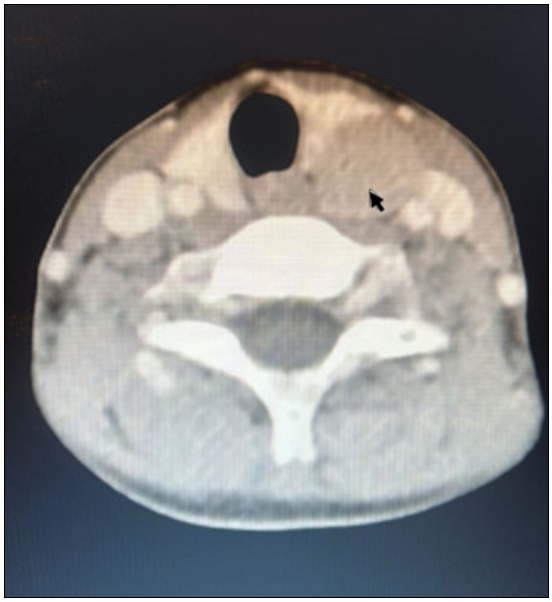

Fig 5: Axial view four-dimensional 4D CT of neck showing well defined hypodense solid lesion arrow posterior to posterolateral surface of left thyroid gland suggestive of parathyroid adenoma

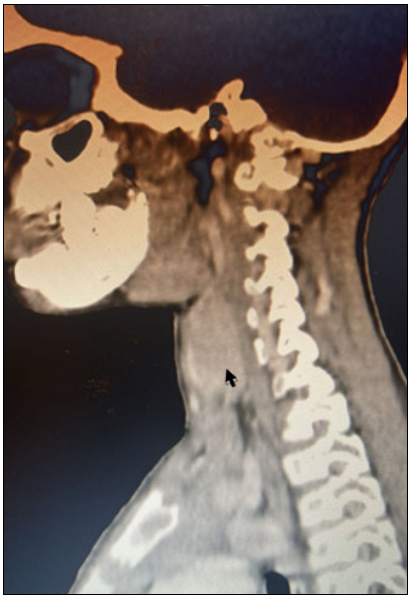

Fig 6: Saggital view of four-dimensional 4D CT of neck showing well defined solid lesion in the tracheo-esophageal groove arrow measuring 2.1× 1.5cm

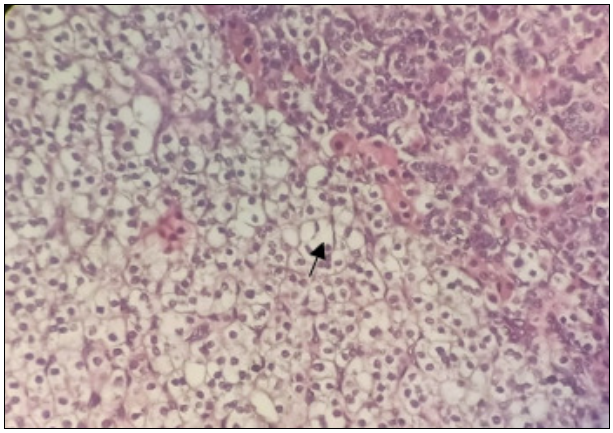

Fig 7: Histopathology of left superior parathyroid gland showing round to oval cells arrow with clear cytoplasm suggestive of chief cell predominant parathyroid adenoma

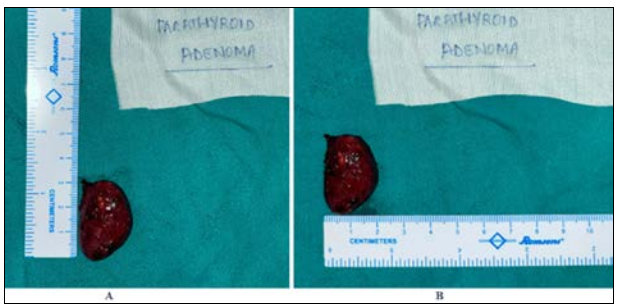

Fig 8: Showing smooth, encapsulated, oval shape of left superior parathyroid gland after resection measuring upto 3×2cm

Table 1: Biocemical profile of the patient during the course of diagnosis and treatment

Parameters	At Presentation	after 4 weeks	Immediate Post-operative	1 week Post-operative
Calcium [mg/dl], [8.5-10.5]	10.2	11.4	6.8	9.3
Phosphorous [mg/dl], [4-6.5]	5.4	4.8	2.12	4.8
Alkaline Phosphatase [U/L], [160-484]	3269	3846	-	626
25 hydroxy cholecalciferol[ng/ml], [30-100]	31	37	33	38
Parathyroid hormone[pg/ml], [8.7-97.6]	2480	3244	154.9	74
TSH [µIU/ml], [0.4-4.2]	3.95	-	-	-
Creatinine [mg/dl]	1.2	0.8	0.8	0.7
Urine Phosphorous [mg/dl], [2.7-9.4]	2.9			-
Urine Calcium [mg/dl]	10.2			-

Discussion

Primary hyperparathyroidism is usually seen in post-menopausal women and it is uncommon in children and adolescents with only 23 cases reported till date [2]. However, in the developing countries with calcium and vitamin D deficiency, primary hyperparathyroidism can be seen in children and adolescents. Literature is sparse on etiology of primary hyperparathyroidism in adolescents and it can be attributed to inactivating calcium-sensing receptor mutation [2]. Other causes of primary hyperparathyroidism can be Multiple Endocrine Neoplasm [MEN-1] syndrome characterized by pituitary adenoma, parathyroid hyperplasia/adenoma and pancreatic tumors. It can also be rarely associated with MEN-2A syndrome [3] characterized by medullary thyroid carcinoma, parathyroid hyperplasia and pheochromacytoma.

Our patient in this study presented with clinical features of short stature, widening of wrist and ankle joints, bilateral genu valgus deformity which may be attributed to the co-existence of vitamin D deficiency with primary hyperparathyroidism [4] mimicking rickets. Literature review by Pitukcheewanout P *et al* [5] showed that Primary hyperparathyroidism in adolescents can present with genu valgus deformity 70%, rachitic rosary 50%, widened wrist and ankle 31.5%, bone/joint pain 37.5% and anorexia 31.25% which can resemble rickets and leads to misdiagnosis.

In a literature review by Pitukcheewanout [5], 13 children had hypercalcemia and only three children were normocalcaemic at presentation who became hypercalcaemic after vitamin D supplementation. In a review by Roizen and Levine [1], it was reported that incidence of hypercalcaemia and hypercalcuria are higher in younger age group compared to adults with primary hyperparathyroidism. In our patient also there was initial normocalcaemia which later developed hypercalcaemia after 4 weeks of vitamin D supplementation which can be attributed to the role of vitamin D in the absorption of calcium from the gastrointestinal tract and reducing renal excretion. Persistently elevated levels of parathormone had deleterious effect on chondrocytes and osteocytes which was not reversed by vitamin D supplementation. There were no gastrointestinal symptoms and other nutrient deficiency excluding malabsorption from the diagnosis.

In the series by Pitukcheewanout [5], 13 out of 14 children had adenoma and only one child had multiglandular disease. All four children had single adenoma in a report by Ganie [6]. The patient in our study had a single adenoma of left superior parathyroid gland which was confirmed by four-dimensional 4D CT followed by histopathological examination and revealed that primary hyperparathyroidism in our patient was due to parathyroid adenoma. Case report by Dutta D [2] also showed that their patient had single adenoma.

Primary hyperparathyroidism in adolescent age can be masked of its classical phenotypical features by presenting as rickets. Hence, in all patients presenting with features of rickets at this age group with no significant improvement after vitamin D supplementation, evaluation should be done to rule out primary hyperparathyroidism which could be due to parathyroid adenoma. Symptoms like bone and joint pain, muscle weakness in our patient were resolved after parathyroidectomy with improvement in appetite. Genu valgus was persisted which required corrective surgery and the overall quality of life of the patient improved.

Conclusion

Primary hyperparathyroidism, though rare in children and adolescents, can present with symptoms mimicking rickets, such as bone deformities and muscle weakness. This case underscores the importance of considering primary hyperparathyroidism in differential diagnoses when patients exhibit rickets-like symptoms that do not improve with conventional vitamin D supplementation. The adolescent patient, initially misdiagnosed with rickets, was ultimately found to have a parathyroid adenoma. Post-surgical intervention led to significant improvements in symptoms, confirming that accurate diagnosis and treatment of primary hyperparathyroidism can markedly enhance quality of life and resolve associated musculoskeletal issues.

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