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General Surgery

PRIMARY HYPERPARATHYROIDISM: A RARE CAUSE OF GENU VARUM DEFORMITY IN ADOLESCENCE

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ABSTRACT Primary hyperparathyroidism is a rare condition in children and results from excessive secretion of parathyroid hormone. The most common cause being a parathyroid adenoma. The presence of limb deformity is atypical with very few cases of genu valgum being described in literature in association with hyperparathyroidism.We present a case of 15year old girl with bilateral genu varum deformity. Patient had hypercalcemia, hypophosphtemia, raised alkaline phosphatase and parathormones with normal renal and thyroid functions. Neck ultrasound done revealed a left inferior parathyroid nodule, skeletal survey showed subperiosteal resorption and osteopenia and MIBI with bone scan confirmed the diagnosis of left inferior parathyroid adenoma. Patient underwent left inferior parathyroidectomy. Following surgery her biochemical parameters including parathormone levels became normal with an initial episode of hypocalcemia(managed with intravenous and oral calcium)in the postoperative period.Bone pain decreased,though the deformity persisted.

KEYWORDS : Hyperparathyroidism, Hypercalcemia, Genu Varum

INTRODUCTION :

Primary hyperparathyroidism is a rare condition in children and results from excessive secretion of parathyroid hormone.(1)(2)The most common cause being parathyroid adenoma in one of the parathyroid glands. A less common cause is multiple endocrine hyperplasia. The incidence of juvenile hyperparathyroidism is 2-5cases/million(3). The common presentations are fatigue, lethargy, headache, nausea, vomiting, abdominal pain,polyuria and excessive sweating. Skeletal manifestations include bonepains and fractures. The presence of limb deformity is atypical and very few cases of genu varum deformity associated with hyperparathyroidism have been described.

CASE REPORT:

A 15year old girl child presented with bilateral genu varum deformity of 4-5months duration,bodyaches and short stature. No significant family history obtained. Lab investigations revealed hypercalcemia, hypophosphatemia, raised alkaline phosphatase and raised parathormones. Renal and thyroid functions were normal. Skeletal survey showed subperiosteal resorption and osteopenia.neck ultrasound revealed left inferior parathyroid nodule and MIBI scan confirmed the diagnosis of left inferior parathyroid adenoma.Patient was initially managed with diuretics and pamidronate,however the response was not good and the patient had to undergo left inferior parathyroidectomy.In the post operative period,the parathormone levels became normal however she developed hypocalcemia for which iv calcium gluconate was given till the serum calcium levels become normal. Patient was discharged on oral calcium on the ninth post operative day .

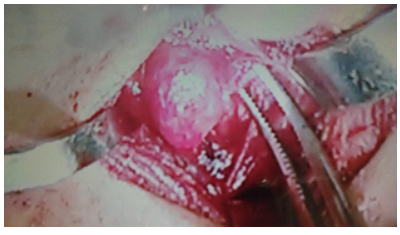

figure 1: Parathyroid adenoma(intra operative picture)

DISCUSSION :

Primary hyperparathyroidism is the result of an intrinsic abnormality of one or more of the parathyroid glands. The abnormal parathyroid gland enlarges and secretes parathormone inappropriately relative to the serum ionised calcium level. Excess circulating parathormone results in increased serum calcium levels. Approximately 85% of patients with primary hyperparathyroidism have single parathyroid gland enlargement;termed as parathyroid adenoma.(4)Less common causes are multiple endocrine neoplasia,parathyroid carcinoma, parathyroid hyperplasia,irradiation to head and neck in childhood etc.

Patients with primary hyperparathyroidism are typically identified incidentally with an elevated total calcium or following routine assessment of bone densitometry. Most of them will have vague constitutional symptoms like fatigue, muscle weakness,depression or memory impairements .Renal and bone involvement are frequently seen in primay hyperparathyroidism and the clinical manifestations include nephrolithiasis,nephrocalcinosis, osteitis fibrosa cystica, pathological fractures,brown tumors,deformity etc.(5)

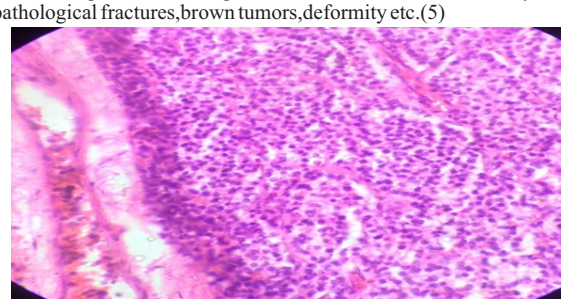

Figure 2:Parathyroid adenoma composed of chief cells and oxyphil cells (microscopic picture)

Bone disease in severe primary hyperparathyroidism (PHPT) is described classically as osteitis fibrosa cystica (OFC). Bone pain, skeletal deformities and pathological fractures are features of OFC. Bone mineral density is usually extremely low in OFC, but it is reversible after surgical cure. The signs and symptoms of severe bone disease include bone pain, pathologic fractures, proximal muscle weakness with hyperreflexia. Bone involvement is typically characterized as salt-and-pepper appearance in the skull, bone erosions and bone resorption of the phalanges, brown tumors and cysts. In the radiography, diffuse demineralization is observed, along with pathological fractures, particularly in the long bones of the extremities(6).

The gold standard for diagnosis of primary hyperparathyroidism is biochemical assessment of parathormone levels & its correlation with serum calcium levels and is supported by radioimaging studies ,bone scans and skeletal survey.Osteoporosis,osteopenia and subperiosteal resorption are the main frequent bony changes described on radiographs.

Parathyroidectomy is the mainstay of treatment.Complications like hypocalcemia,hoarseness of voice ,damage to recurrent laryngeal nerve(<1%),hemorrhage,persistent(5%) or recurrent hyperparathyroidism, permanent hypoparathyroidism(0.5%) may

occur in the post operative period.(5)

CONCLUSION :

Parathyroid adenoma presenting with bilateral genu varum deformity is rare in children. The presence of hypercalcemia in the presence of deformities points to the possibility of primary hyperparathyroidism. Parathyroid adenoma in one of the parathyroid glands is the commonest etiology. Surgical excision of the parathyroid adenoma may prevent deformities with symptomatic improvements.

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