

OSTEITIS FIBROSA CYSTICA IN A YOUNG MALE

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ABSTRACT

The classic skeletal manifestation of advanced primary hyperparathyroidism is called as Osteitis Fibrosa Cystica (OFC). The paradigm has now shifted from identifying primary hyperparathyroidism to asymptomatic primary hyperparathyroidism owing to the wide spread use of measuring serum calcium. We describe a classical feature of advanced primary hyperparathyroidism due to a parathyroid adenoma and its successful treatment.

Key words : osteitis fibrosa cystica, hyperparathyroidism, brown tumour, APLAR Grand Round Case

INTRODUCTION

Osteitis fibrosa cystica (synonyms- osteitis fibrosa, osteodystrophia fibrosa, and Von Recklinghausen's disease of bone) was first reported by von Recklinghausen in 1891. It is a disorder of skeletal system caused by hyperparathyroidism due to over functioning parathyroid gland. It is not a true neoplasm but rather a reactive osteolytic lesion of bone and may mimic other diseases such as giant cell tumors, multiple bone metastasis or multiple myeloma¹. Women are thrice affected than men. Plain radiographs show skull resorption with “ground glass”/“salt and pepper” appearance. The earliest X-ray features are seen in the fingers. The cysts are lined by osteoclasts and sometimes blood pigments, hence called brown tumours¹. These cysts can be identified with Sestamibi nuclear imaging

CASE REPORT

A 31 years male, a manual labourer by occupation, presented to our Rheumatology OPD for insidious onset, progressively worsening mid and low back pain. The pain was present throughout the day, aggravated by bending and lifting heavy weight. There was no radiation of pain. There was no limitation of movements, but had to do his work with moderate difficulty. He had no fever, loss of appetite or loss of weight. There was no preceding diarrhea, dysuria, red eyes, skin lesions or foot pain. He also had pain over the small joints of both hands, which was not associated with swelling or stiffness. On further questioning, he also had pain in the right thigh. He reported a significant fatigue, which interfered with his daily activities. He had mild

difficulty in getting up from squatting and climbing stairs. He had polyuria. There was no swelling of legs, abdominal distension or puffiness of face. He also had abdominal pain, which was of burning type, localized to the epigastrium with occasional nausea, vomiting and belching.

On examination he was not pale. He didn't have clubbing, lymphadenopathy, puffiness of face or pedal edema. His blood pressure was 150/90mmHg in the right upper limb with no disparity in the other limbs. Other vitals were normal. There was no goiter.

Musculoskeletal examination showed tenderness over the metacarpophalangeal joints (squeeze test was positive), proximal and distal interphalangeal joints. There was no palpable synovial thickening or effusion in the small joints of the hand. Medium and large joint examination was clinically normal.

Patrick's test and Gaenslen's test were negative. He had no skin lesions, enthesitis or dactylitis. Spine examination showed tenderness along the lower thoracic spine (T9-T12). There was no gibbus. He also had a diffuse bone pain.

Systemic examination showed epigastric tenderness. Neurological examination showed a mild proximal muscle weakness with a power of 4/5, with preserved reflexes and intact sensory system. All other systems were clinically normal.

On laboratory analysis, complete blood count was normal. Erythrocyte sedimentation rate (ESR) was 32mm/hour. Serum calcium level was 11.2 mg/dl (normal 8.4-10.7 mg/dl), serum albumin level was 5 g/dl (normal 3.4-4.8 g/dl), and corrected calcium was 10.4mg/dl. Serum alkaline phosphatase level was 720 IU/l (normal 50-240 IU/l), serum acid phosphatase level was 10.16 IU/l (normal 0-5.5 IU/l), serum parathyroid hormone (intact) level was 335 pg/ml (normal 7-53 pg/ml), and vitamin D3 (1, 25-dihydroxy cholecalciferol) was 32 pg/ml (normal 25-45 pg/ml). Renal functions were normal. Thyroid functions were normal.

Ultrasound of neck showed a well defined homogenous hypo echoic solid soft tissue lesion in relation to the posterior aspect of inferior pole of left lobe of thyroid.

X ray of hands showed subperiosteal bone resorption along the radial aspects of the middle phalanges (fig 1) and digits (fig 2). X ray of thoracolumbar spine showed alternating areas of radio dense and radiolucent areas (ruger jersey spine) along with nephrocalcinosis (fig 3). X-ray of skull showed salt and pepper appearance with well defined osteolytic lesions (fig 4). X ray of right thigh showed well defined lytic lesion in lower third of shaft of femur (fig 5).

A diagnosis of Osteitis fibrosa cystica due to primary hyperparathyroidism owing to a parathyroid adenoma was made and patient was referred to endocrine surgery where he underwent parathyroidectomy.

DISCUSSION

Osteitis fibrosa cystica is more commonly seen in women. It occurs more in the 5th and 6th decades. If it occurs earlier (especially first decade), hereditary causes—multiple endocrine neoplasia type I/IIa/IIb has to be ruled out.

Primary hyperparathyroidism results from excessive secretion of parathyroid hormone (PTH). It can be caused by a solitary adenoma in 80% patients³, parathyroid hyperplasia in 15%, multiple adenomas in 5%, and parathyroid carcinoma in less than 5% of patients². 80–85% of cases of hyperparathyroidism are due to parathyroid adenoma. The increase in parathormone stimulates the activity of osteoclasts that breaks down the bone. Decreased bone mineral density and nephrolithiasis are the major sequelae of hyperparathyroidism⁴. The manifestations of this disease are the consequences of both the general softening of the bones and the excess calcium in the blood. They include bone fractures, nephrocalcinosis, nausea, peptic ulcers, constipation, acute pancreatitis, appetite loss, and weight loss—“bones, stones, abdominal groans and psychic overtones”. Neuropsychiatric manifestations include proximal myopathy, weakness and easy fatigability, depression, inability to concentrate, and subtle memory deficits which may not be noted by the patient. Hypertension, bradycardia, shortened QT interval, and left ventricular hypertrophy are the predominant cardiovascular manifestations.

DIAGNOSIS

Earliest and most pathognomonic sign of hyperparathyroidism is subperiosteal bone resorption. This commonly occurs in the middle phalanges of the index and middle fingers, primarily on the radial aspect. Other sites include the phalangeal tufts, the medial aspect of the tibia, the humerus, the lamina dura around the teeth, the femur, and the distal clavicle. In the skull, areas of decreased radiopacity along with sclerotic areas, result in a classical salt-and-pepper skull. Brown tumors are well-circumscribed lytic bone lesions which represent the osteoclastic resorption of an area of bone with subsequent fibrous replacement. The serum calcium, Parathormone, and alkaline phosphatase are usually elevated. Ultrasonography of the neck localizes parathyroid tumors.

TREATMENT

Parathyroidectomy is the only effective therapy for osteitis fibrosa cystica in hyperparathyroidism⁵.

PECULIARITIES OF THIS CASE

1. Male preponderance
2. Younger age of onset without MEN syndrome.
3. Rugger jersey spine, nephrocalcinosis and brown tumours are more common in secondary rather than primary hyperparathyroidism. Ours is a case of primary hyperparathyroidism due to parathyroid adenoma.
4. Overt radiological bone disease (osteitis fibrosa cystica) is almost never seen⁶ due to earlier detection and effective management.

CONCLUSION

Osteitis fibrosa cystica can mimic many diseases. A simple serum calcium estimation can give a clue to the diagnosis. A diagnostic confusion arises when there are multiple lytic lesions

involving different areas of the skeleton. In case of hypercalcemia and radiographic evidence of multiple lytic lesions, primary hyperparathyroidism should always be kept in differential diagnosis.

Fig 1. X- ray of hands showed show subperiosteal bone resorption along the radial aspects of the middle phalanges



Fig 2. X- ray of hands showed show subperiosteal bone resorption of digits(acro osteolysis)



Fig 3. X- ray of thoracolumbar spine showed alternating areas of radio dense and radiolucent areas (ruger jersey spine) along with nephrocalcinosis



Fig 4. X-ray of skull showed salt and pepper appearance with well defined osteolytic lesions



Fig 5. X- ray of right thigh showed well defined lytic lesion in lower third of shaft of femur



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