CASE REPORT

Hyper Parathyroidisim Jaw Tumor Syndrome: A Rare Condition of Incongruous Features

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ABSTRACT

BACKGROUND: ***Hyperparathyroidism-Jaw Tumor (HPT-JT) syndrome is a rare genetic disorder bearing both a germline and a somatic CDC73 mutation (formerly known as HRPT2), which has been mapped to chromosome 1q25-q31. The association of jaw ossifying fibroma with primary hyperparathyroidisim (PHPT) is typical of HPT-JT. It may also include cystic and neoplastic renal abnormalities and uterine tumors.***
CASE DETAILS: Here, we report a case of HPT-JT with an initial presentation of declination in reproductive fitness. Extensive literature search and thorough investigation helped us parturitate the underlying syndrome, thereby predictively improving the prognosis.
CONCLUSION: The features of HPT-JT are clinically difficult to ascertain because the parathyroid disease, ossifying fibroma in the jaw and other abnormalities, often occurs asynchronously and may be diagnosed and treated separately.
KEYWORDS: Hyperparathyroidism, HPT-JT, Ossifying fibroma, Syndrome

INTRODUCTION

Jackson was the first to describe Hyperparathyroidism-Jaw Tumor (HPT-JT) as one of the rare and relatively unknown hyperparathyroidism-related syndromes, with unanticipated clinical features (1). It is an autosomal dominant disorder with incomplete penetrance and variable expression. It includes parathyroid adenomas, ossifying fibroma of the jaw, uterine tumor in females and renal diseases such as hamartomas, polycystic disease and Wilms tumors or adenocarcinoma. Diagnosis of HPT-JT is important because of its genetic involvement and 24% malignant transformation (2).

CASE PRESENTATION

A 23-year-old female reported to our Department of Oral Medicine and Radiology, Sree Mookambika Institute of Dental Sciences, Kulasekharam, Kanyakumari District-629161,

around 2 years ago. She presented with, a slow growing, painless swelling in her lower right back tooth region since a year. She gave a history of a similar swelling on the left, which was surgically removed eight years back, but further details of the same were unavailable. Her medical history revealed multiple miscarriages with inability to sustain pregnancy.

On examination, the swelling was extending from 44 to 46 in the vestibular region (Figure 1), and it was bony hard, smooth and tender on palpation. The teeth in the region responded positively to the electric vitality test. Based on the site, age, sex and clinical examination, the lesion was provisionally diagnosed as, central giant cell granuloma.

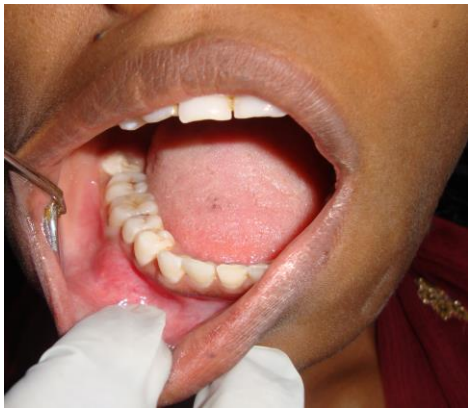

Figure 1*: Intra Oral Photograph showing diffuse swelling in the right side posterior region of mandible*

Orthopantamograph revealed a multilocular radiolucency on the right side body of the mandible and an Ill-defined radiolucent area on the left (Figure 2). Computed Tomography revealed its anatomical extension and confirmed its features (Figure 3). The radiographic impression of the lesion of interest was central ossifying fibroma, and the differential diagnosis was given as Central Giant cell granuloma and Ameloblastoma. The remnant lucency on the left mandibular posterior region was regarded as a surgical scar/defect.

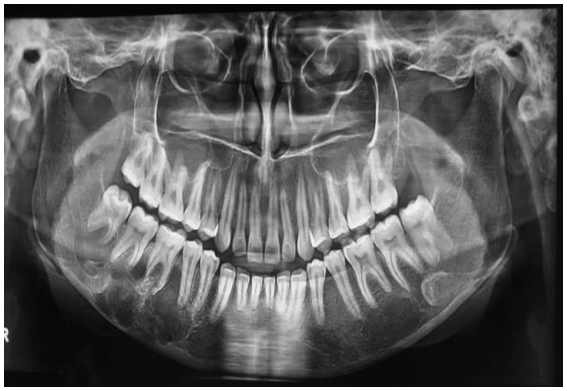

Figure 2: Orthopantomogram revealing multilocular radiolucency in the right side body of mandible

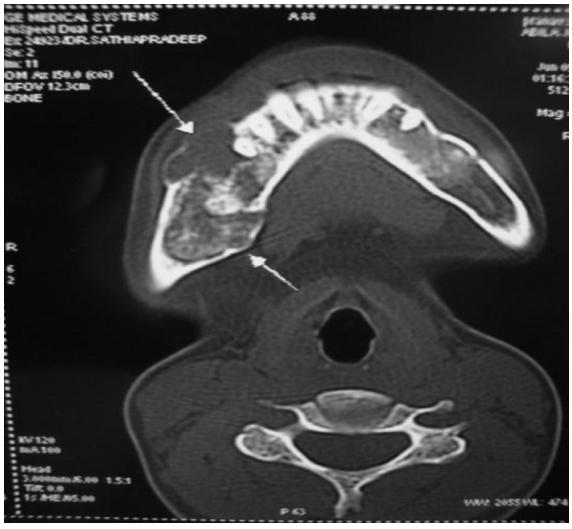

Figure 3*: Computed Tomography of mandible (Axial view) revealing buccal and lingual plate expansion*

Incisional biopsy was done, which histopathologicaly revealed it as ossifying fibroma. Serological Biochemistry revealed rise in serum calcium (14.2mg/dl) and alkaline phosphatase (369IU/L) along with low phosphorus level (2mg/dl). Accordingly, Parathyroid Hormone (PTH) level was estimated, which revealed its elevation (135pg/ml) and confirmed primary hyperparathyroidism. In order to exclude Multiple Endocrine Neoplasia, other investigations such as routine blood and urine, random blood sugar, thyroid function test, liver function test and prolactin level were estimated and found to be within normal limits.

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Ultrasonography of the neck revealed a well-defined oval hypo echoic mass (13×9mm) posterior to the left lobe of thyroid gland showing minimal vascularity within. This was further confirmed by MRI and Tc-99m parathyroid imaging (Figure 4). Renal Ultrasonography revealed three calculi of 0.5cms in the calyx.

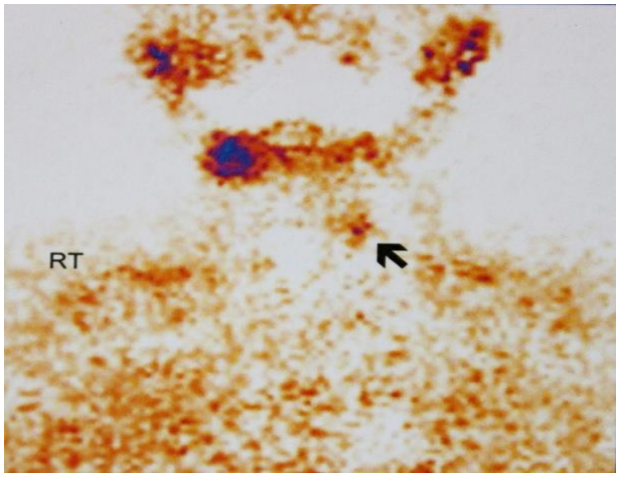

Figure 4*: Scintigraphy of Head and Neck revealing hot spot in the left parathyroid gland*

Left parathyroidectomy was executed, which histopathalogically confirmed parathyroid adenoma. Post-surgical biochemical evaluation revealed a drastic decline in the level of serum calcium (8.8mg/dl), alkaline phosphatase (225IU/L), parathyroid hormone (75.2pg/ml) and rise in level of phosphorus (3.8mg/dl). The mandibular lesion was surgically enucleated (Figure 5), which histopathologically confirmed central ossifying fibroma. Reconstruction with Iliac bone graft and metal plating were performed.

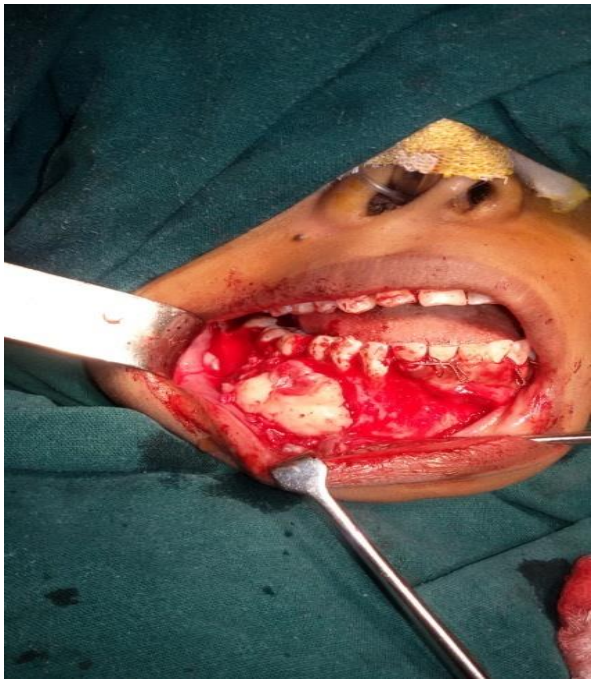

Figure 5*: Surgical enucleation of the mandibular lesion*

We decided to further explore the true underlying etiology and began to consider HPT-JT syndrome as the possible explanation. Based on this assumption, we advised sonographic investigation of the abdomen, which revealed adenomyosis (1.6 cms) of the enlarged uterus.

On correlating the presence of ossifying fibroma of the jaw and uterine tumor with primary hyperparathyroidism, without any other features of complex syndromes associated with hyperparathyroidism, we arrived at a confirmatory diagnosis of HPT-JT syndrome. Gene mapping for confirmation was recommended. However, the exorbitant cost involved and the fact that it did not have a bearing on the treatment and the prognosis was a deterrent to the patient. No signs of recurrence, malignant changes or any abnormal serum biochemical levels were noted in her follow up (Figure 6). The patient was further referred to the Department of Obstetrics and Gynecology for management of the uterine condition and to the Department of Nephrology for management of the renal calculi.

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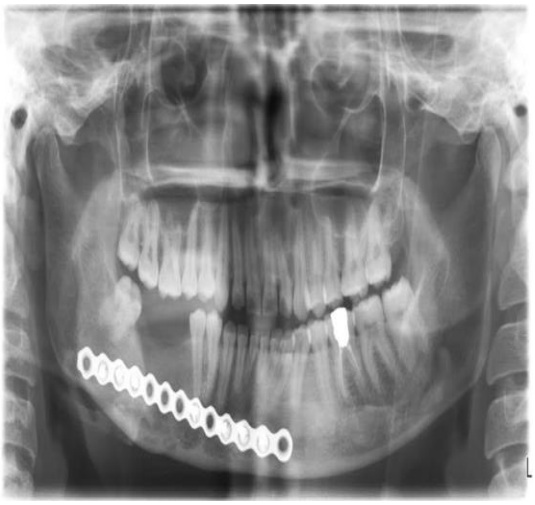

Figure 6*: Post operative Radiograph (OPG) revealing bone formation in the surgical site*

DISCUSSION

HPT-JT affects at least three distinct tissues: parathyroid glands, gnathic skeleton and the kidney (3). PHPT is the first manifestation of the disease in more than 95% of HPT-JT patient as solitary parathyroid adenoma (4). Uniquely, in our case, declination in reproductive fitness was the first clinical manifestation followed by jaw tumor.

The jaw tumors in HPT-JT are different from brown tumors observed in some patients with primary HPT, and such jaw tumors do not resolve after parathyroidectomy. Indeed, they are ossifying fibroma of the jaw, which is an important distinguishing feature of HPT-JT (3).

Complete excision of the lesion and reconstruction should be considered when necessary, as it has a tendency to recur with incomplete resection (5). Ideally, in our case, Iliac bone grafting and surgical plating were done after complete resection inorder to esthetically re-contour the jaw.

Uterine tumour appears to be common in HPT-JT; in such cases, reproductive fitness declines for upto 75% of women (6). In our patient, adenomyosis was found in the Ultrasonic investigation and explained the history of multiple miscarriages with inability to sustain pregnancy. Excessive rise in PTH is the Biochemical hallmark of PHPT, while hypercalcemia is considered the clue for diagnosing PHPT (7). These biochemical evaluations paved the way for diagnosing the hidden abnormalities in our patient.

To be more precise, all the patients with clinical and biochemical evidence of hyperparathyroidism should be subjected to a pre-operative Tc-99m Sestamibi imaging (8). In our patient, various advanced imaging modalities including Tc-99m Sestamibi were utilized to confirm the diagnosis.

The absolute treatment for Parathyroid Adenoma is parathyroidectomy. However, in some patients, for whom surgery is contraindicated or who refuse surgical procedure, can be treated medically with Cinacalcet hydrochloride, Conjugated estrogen + medroxyprogesterone or Alendronate to safeguard the bone (9). Even after the surgical correction, both Ossifying Fibroma and parathyroid adenoma carry a risk of recurrences; this necessitates the need for long term follow-up (10).

Collaboration between healthcare professionals and an interdisciplinary approach to the patient issues will lead to early diagnosis, comprehensive treatment and increased quality of patients' lives, especially in cases like HPT-JT. This article is an effort to highlight the importance of thorough investigations and a reminder to all concerned, to be aware of the possibility of the HPT-JT syndrome in adolescents and young adults.

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