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Case Report

Renal osteodystrophy and brown tumours of the jaw associated with tertiary hyperparathyroidism in the context of cystinosis: a case report

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ABSTRACT

Chronic kidney disease is one of the main manifestations of cystinosis that may lead to hyperparathyroidism. Although head and neck involvement in hyperparathyroidism is rare, lesions can occur in the jaws and cause serious functional handicap. We presented the case of a 24-year-old woman with congenital cystinosis and chronic renal failure complaining from symptomatic expansion of the jawbones. Medical imaging showed radiolucent lesions particularly in anterior hard palate and body of mandible. A diagnosis of renal osteodystrophy and brown tumours related to tertiary hyperparathyroidism was established. Following a subtotal parathyroidectomy and stabilization of parathyroid hormone levels, a conservative mandibular osteoplasty was undertaken to increase the available space for tongue movements. Although non-surgical management remains the primary modality for the treatment of brown tumours, adjunctive surgical approach may be considered in cases of severe functional impairment. It is imperative that underlying systemic imbalances be addressed prior to any maxillofacial intervention to prevent relapse.

Keywords: Renal osteodystrophy, Chronic kidney disease, Osteitis fibrosa cistica, Brown tumour, Tertiary hyperparathyroidism, Cystinosis

INTRODUCTION

Cystinosis is a rare genetic disorder in which the accumulation of cystine in lysosomes leads to ocular and renal lesions. Left untreated, a chronic kidney disease (CKD) is inevitable.[1] Hyperparathyroidism (HPT) is defined as an overproduction of parathyroid hormone. In its primary form, it involves a *de novo* phenomenon whereas in its secondary type, it is a physiological response to metabolic stimuli, typically in the context of CKD. Tertiary HPT is a result of a prolonged state of secondary HPT such as a long-standing CKD, and is characterized by semi-autonomous parathyroid hormone over-secretion.[2]

Osteitis fibrosa cystica (OFC), also known as brown tumours of hyperparathyroidism, are well-defined radiolucent lesions rarely involving head and neck region.[3] Neoplastic lesions can also be seen in HPT as hyperparathyroidism-jaw tumour syndrome (HPT-JS). In this rare autosomal dominant disorder, a history of familial parathyroid adenomas, ossifying fibroma of the jaws and renal lesions are significant.[4]

CASE REPORT

A 24-year-old white female known for congenital cystinosis was referred for assessment of painful progressive maxillary and mandibular enlargement for 4 months. She was under haemodialysis treatment three times per week for the past 4 years because of CKD. The patient had a renal transplant rejection at the age of 7. Furthermore, her medical history showed anaemia secondary to CKD, hypertension, hypothyroidism, splenomegaly and migraines. No family history of HPT, endocrine neoplasia, jaw tumours or kidney disease had been documented.

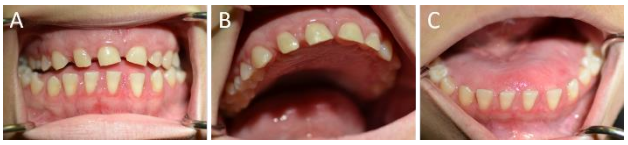

Figure 1: Clinical photographs showing the anterior open-bite and dental displacement in maximum intercuspation (A) as well as notable expansion on hard palate (B) and lingual surface of mandible (C).

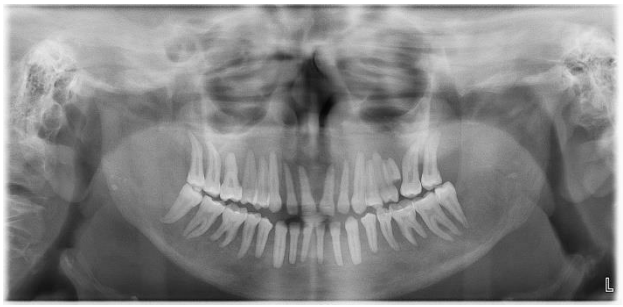

Figure 2: Panoramic radiography showing ground glass aspect of the maxillary and mandibular bone and absence of lamina dura around radicular portion of the whole dentition.

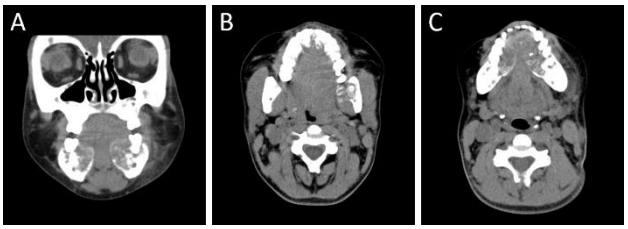

Figure 3: Coronal (A) and axial (B,C) sections of a cranio-facial computed tomography scan without radiocontrast agent showing hypodense intraosseous lesions on medial surface of posterior mandible (A), anterior hard palate and left mandibular ramus (B) in addition to lingual aspect of mandibular body (C).

Short stature and insufficient vertical jaw growth with labial incompetence were noted in the extraoral examination. Intraorally, a massive cortical bone expansion was observed mainly on hard palate and lingual cortical surface of mandible bilaterally (Figure 1).

The blood analysis history showed high levels of parathyroid hormone: 360 pmol/l comparing to the normal range of 1.09 to 6.87 pmol/l. A panoramic radiography showed diffused ground glass appearance of the entirety of maxilla and mandible (Figure 2). No focal lesion was noted. Several rounded apices suggested a decelerated dental apex formation. No lamina dura was identifiable regarding the periodontium of the entire dentition.

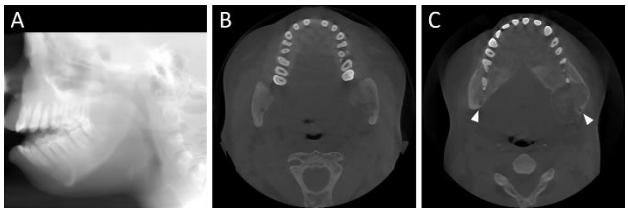

Figure 4: Antero-posterior view (A) of the scanned area in cone beam computed tomography showing the patient's profile and lack of vertical growth of the jaws. Axial sections confirm the expansion of the hard palate and left mandibular ramus (B) and presence of a heterogenous lesion in left mandibular ramus (C) displacing left mandibular canal comparing to the right side (arrow heads).

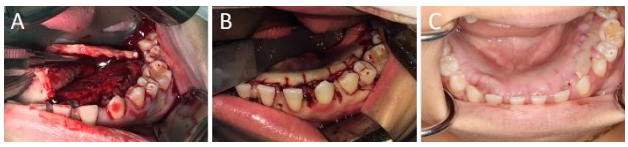

Figure 5: Clinical photographs during mandibular osteoplasty surgery (A), immediately after the end of the operation (B) and at the post-operative follow-up 2 weeks later (C) showing notable improvement of the available space for mobile tongue.

A cranio-facial computed tomography (CT) scan without contrast-because of the patient's renal function-revealed the presence of hypodense lesions with large areas of cortical expansion (Figure 3). A cone beam CT that was performed later also confirmed the generalized reduction of cortical bone, maxillary sinus atrophy and presence of a heterogenous lesion in the left mandibular ramus displacing left mandibular canal (Figure 4).

A working diagnosis of renal osteodystrophy and brown tumours in relation with chronic hyperparathyroidism was established. Nephrology, otolaryngology as well as endocrinology consultations were planned in order to address the hyperparathyroidism. The pathology results after a subtotal parathyroidectomy confirmed the clinical suspicion of parathyroid hyperplasia.

Based on the complaints of the patient regarding the available space for tongue movements, a surgical

approach to correct the bony deformity was discussed. Once the parathyroid hormone levels stabilized, a mandibular osteoplasty was planned. A lingual mucoperiosteal flap was raised after sulcular incision around mandibular teeth and lesion debulking was performed with surgical round bur and manual curette (Figure 5A). Primary closure was obtained via simple sutures chromic gut 3-0 for each interproximal papilla (Figure 5B). Patient was discharged from hospital after 2 days with no complications. A 2-week follow-up showed notable improvement of the tongue movements owing to the increase of the free space on the floor of the mouth (Figure 5C).

DISCUSSION

OFC are non-neoplastic reactive lesions. However, with their multifocal osteolytic involvements they can mimic other entities such as metastatic bone lesions and multiple myeloma. A thorough analysis is needed to avoid unnecessary surgical interventions due to misdiagnosis.[5]

In primary HPT, the main option of treatment is parathyroidectomy especially in symptomatic patients. In contrast, the correction of hypocalcaemia and hyperphosphataemia is the main goal in the treatment and prevention of renal osteodystrophy by addressing the underlying metabolic imbalance in HPT. This can be achieved mainly via calcium and phosphate binders and calcitriol.[6,7]

OFC usually regresses spontaneously after the stabilization of parathyroid hormone, but surgical resection can be beneficial in cases of persistent lesions or large deformities. If adjunctive bone surgery is planned, a controlled parathyroid hormone levels should be attained prior to intervention since bone healing would be compromised otherwise.[8-10]

CONCLUSION

This case report provides valuable insight into the presentation of brown tumours and renal osteodystrophy in patients with cystinosis and chronic kidney disease. It highlights the importance of accurate diagnosis, control of parathyroid hormone levels, and consideration of surgical intervention for severe functional impairment. The report contributes to the growing body of knowledge on renal osteodystrophy and brown tumours related to tertiary hyperparathyroidism and the role of interdisciplinary management in such cases.

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