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Delayed Diagnosis of Severe Osteomalacia in a Patient with RTA

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INTRODUCTION

A 37-year-old lady was admitted to the medical ward for complaints of worsening body and muscle weakness and generalized bony pain for the past 4 years following a minor motor vehicle accident causing significant debility and disability. She was diagnosed with RTA in 1999 complicated by nephrocalcinosis however had defaulted follow up and medications. Her sister was also diagnosed with RTA albeit with milder symptoms.

CASE

Clinical examination revealed generalized proximal muscle weakness with no focal neurological signs or cranial nerve deficits. Biochemical investigations revealed normal anion metabolic acidosis, hypokalemia (K 2.5 mmol/l), hypophosphatemia (PO4 0.49 mmol/L), normal corrected calcium of 2.20 mmol/L, a high ALP level of 549 IU/L and a normal PTH level of 19.6 pg/ml. The Fractional excretion of phosphate was elevated at 21.09% suggestive of urinary phosphate wasting with no hypercalciuria. Vitamin D level (25 OH Vitamin D) was low at 10 nmol/L. Radiographs of the extremities and pelvis done showed multiple looser zones at the right scapula, pelvic rami and right neck of femur. A BMD scan revealed a Total T score of -4.0 and -3.5 at the hip and spine respectively. Nerve conduction study done during the admission was suggestive of chronic myopathy of systemic disease.

A diagnosis of severe osteomalacia secondary to RTA with coexistent Vitamin D deficiency was made. The patient was started on high dose Vitamin D, Shohl's solution with potassium supplementation and was referred for rehabilitation and physiotherapy.

CONCLUSION

Phosphate levels normalized and repeated Vitamin D level improved to 29 nmol/L and the patients pain and muscle power has significantly improved since then. A BMD scan is planned in a year to reassess response to therapy.

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Triple Synchronous Tumors presenting as Right Nasolabial Basal Cell Carcinoma, Papillary Thyroid Carcinoma and Prolactinoma: A Rare Case Report

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INTRODUCTION

A 57-year-old female presented with triple synchronous tumors with a rare combination of basal cell carcinoma (BCC) of the right nasolabial area, papillary thyroid carcinoma (PTC) and prolactinoma. The case initially presented a diagnostic dilemma in approaching multiple tumors from different endocrine organs. On work up, serum prolactin was elevated and an enhancing thyroid mass on neck CT scan and a pituitary mass on cranial MRI were noted. Bromocriptine was given and she underwent total thyroidectomy and wide excision of the right nasolabial BCC. On follow up, repeat serum prolactin decreased back to normal from baseline. This case report aims to provide an organized approach to multiple tumors involving endocrine organs and to reconcile the challenge in ruling out metastasis and syndromic disorders such as Multiple Endocrine Neoplasia. This paper also aims to highlight the need for a multidisciplinary team in the management of multiple tumors.

CASE

After extensive literature review, this is the first documented case of triple synchronous tumors with a combination of BCC of the right nasolabial area, PTC and prolactinoma in local, national and international studies. Double primary tumors are rare with a prevalence rate of 3-5%. Triple primary tumors are even rarer having an occurrence of 0.5%.

CONCLUSION

Accurate knowledge on disease prevalence, pathophysiology and symptomatology correlated with guided history taking and astute physical examination is very important in rare medical conditions and complicated cases. Extensive work up based on sound clinical knowledge and judgement aids also in answering clinical questions encountered in a diagnostic dilemma.