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DIFFERENTIAL DIAGNOSIS FOR OSTEOPOROSIS: AN ONCOGENIC HYPOPHOSPHATEMIC OSTEOMALACIA DIAGNOSIS CASE REPORT

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BACKGROUND

Oncogenic hypophosphatemic osteomalacia (OHO) is a paraneoplastic disorder, in which there's a hypersecretion of phosphatonin – fibroblast growth factor 23 (FGF-23) – by a mesenchymal tumor that leads to urinary phosphate wasting. The tumor-induced osteomalacia (TIO) is typically small and benign. Originally, FGF-23 is produced under physiological conditions by osteocytes to regulate the phosphorus cycle. When overexpressed, this substance causes downregulation of the renal phosphate reabsorption transporter and decreased levels of 1,25 dihydroxyvitamin D [1,25(OH)₂D] due to 1- α -hydroxylase inhibition, resulting in hypophosphatemia, hyperphosphaturia and inappropriate normal/low plasma 1,25(OH)₂D concentration. Besides that, both calcium and parathyroid hormone levels are normal. The prevalence of the OHO is unknown (approximately 400 cases have been reported in the literature). Main clinical features of this disease are muscle weakness, bone pain, pathological fractures or rickets. Diagnosis is challenging and consists of detecting TIO, although it is not always possible. Whole body imaging is usually needed, usually PET CT is performed for screening and magnetic resonance imaging (MRI) to confirm diagnosis. Surgical resection of the tumor is a curative treatment and full recovery is expected.

CASE REPORT

Six years ago, a 75-year-old woman presented with chronic ascending paresthesia and shock-like lower limbs pain. Two years later, she developed generalized weakness, suffering several falls over the time. Due to persistent back pain, a spine MRI was performed and found multiple vertebral fractures (D11 and L1 to S1). A couple of years later, once she also complained of pain in her right ankle, other MRI was made, confirming a new fracture. Laboratory findings revealed no sign of malabsorption or malignant diseases at the time, but showed hypophosphatemia (1.5 mg/dL) and decreased phosphorus tubular resorption capacity (66%). Her bone density test that was normal at the symptoms onset, deteriorated quickly revealing osteoporosis. The hypothesis of OHO was established and confirmed by high levels of FGF-23 (2605). Patient underwent cancer screening and positron emission tomography–computed tomography (PET-CT) detected suspicious accumulation in the thyroid and left femur. Bone biopsy showed poorly differentiated neoplasm and thyroid biopsy was suggestive for Hürthle cell neoplasm, not being possible to identify primary site. She underwent chemotherapy, but developed osteonecrosis of the jaw, requiring suspension of treatment. Patient is currently in symptomatic treatment with phosphate solution.

CONCLUSION

Oncogenic hypophosphatemic osteomalacia should be considered as a differential diagnosis of osteoporosis when associated with rapid progression, hypophosphatemia, muscle weakness and multiple fractures.