

Tumor-induced osteomalacia due to a recurrent mesenchymal tumor overexpressing several growth factor receptors

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Summary

Tumor-induced osteomalacia (TIO) is a rare paraneoplastic syndrome caused primarily by benign mesenchymal tumors. These tumors typically follow a benign clinical course and local recurrence occurs in <5% of cases. We investigated a 49-year-old man with a recurrent mesenchymal phosphaturic tumor showing no signs of malignancy. The patient suffered from chronic muscle weakness, myalgia and cramps. His medical record included the diagnosis of oncogenic osteomalacia, for which he was submitted to tumor resection in the left leg three times before. Laboratory examination showed hypophosphatemia, hyperphosphaturia and an elevated serum FGF23 level. A radical surgical approach (amputation) was advised, however, complete biochemical and clinical remission was not reached. Molecular analysis of the tumor cells demonstrated overexpression of growth factor receptors implicated in tumor angiogenesis and metastatic potential (platelet derived growth factor type A (PDGFRA), PDGFRB and vascular endothelial growth factor receptor) together with increased expression of FGF23, x-linked-phosphate-regulating endopeptidase and KLOTHO. TIO is usually associated with benign phosphaturic tumors and, when identified, resection of the tumor leads to complete remission in the majority of cases. The underlying pathophysiology of recurrences in these tumors is not known. This is the first report showing increased expression of growth factor receptors in a locally aggressive but histopathologically benign phosphaturic mesenchymal tumor.

Learning points:

- TIO is usually associated with benign soft tissue or bone neoplasms of mesenchymal origin.
- These tumors typically follow a benign clinical course and even in the rare malignant cases local recurrence occurs in <5%.
- Successful identification and removal of the tumor leads to full recovery in the majority of cases.