
Proceedings Paper:
Schini, M. orcid.org/0000-0003-2204-2095 and Eastell, R. orcid.org/0000-0002-0323-3366 (2018) Adult Hypophosphatasia : a disease where the clinical complications could be avoided by careful evaluation of patients. In: Osteoporosis International. Osteoporosis Conference 2018, 02-04 Dec 2018, Birmingham, UK. Springer , pp. 632-633.

https://doi.org/10.1007/s00198-018-4738-8

ADULT HYPOPHOSPHATASIA: A DISEASE WHERE THE CLINICAL COMPLICATIONS COULD BE AVOIDED BY CAREFUL EVALUATION OF PATIENTS

Background: Hypophosphatasia (HPP) is a rare disease that has to be excluded before starting treatment for osteoporosis; HPP patients are more prone to develop atypical femur fractures (AFF) from bisphosphonates.
Clinical presentation: A 47-year old woman was referred for bone mineral density (BMD) because of a family history of osteoporosis. Investigations were performed because BMD was below the average for age and the results were reported as normal. A year later, she was prescribed on alendronate. At the age of 54, she sustained two metatarsal fractures. The following year she started noticing odd sensations at her hips and at the age of 56, she tripped and sustained a complete left and an incomplete right femur fracture (considered to be AFFs due to bisphosphonate use).
The patient was then referred for further evaluation. Her past medical history included loss of upper teeth at the age of 20. Her family history included metatarsal fractures in her mother.
Investigations: Investigations were performed three months after the fractures and showed the following: ALP 42 IU/L (30-130), P1NP 86 μg/L (30-78), bone ALP 12 IU/L (9-28). Due to the medical history and the low-normal bone ALP, PLP was measured and was 190 nmol/L (40-100).
Management: The diagnosis of HPP was established and the patient will be sent for genetic testing. The femur fractures were treated surgically with intramedullary nails and 2 months later there was evidence of callus formation.
Conclusion: This patient's ALP was normal on two occasions and the diagnosis of HPP was established after she sustained the fractures. Previous measurements of ALP performed on this patient were low but were ignored. The learning point from this study is that patients considered for treatment with bisphosphonates should be evaluated thoroughly and previously performed investigations should be taken into account.