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### Bilateral Atypical Femur Fractures as a Presenting Manifestation of Unrecognized Hypophosphatasia

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## Learning Objective

- Review hypophosphatasia and describe its atypical presentation in an adult male.

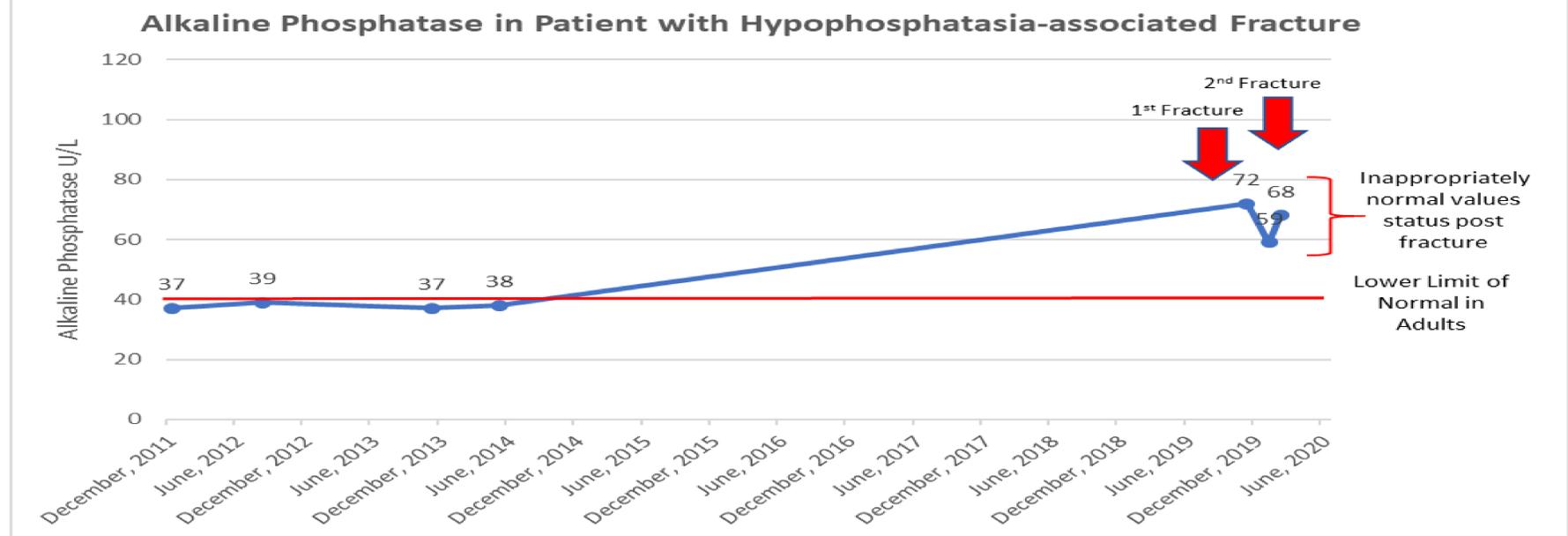
## Introduction

- Hypophosphatasia (HPP) is a rare inherited disorder of the ALPL gene, which leads to decreased alkaline phosphatase (ALP) activity.
- Though severe symptoms are seen in childhood, adult HPP is milder and presents with nonspecific symptoms and thereby is often undiagnosed.
- Hallmarks of adult HPP include decreased ALP, non-traumatic fractures or pseudo-fractures, and premature loss of teeth.

## Case Presentation

- A 56 year old previously healthy man presented to Bone & Mineral Clinic for evaluation of bilateral atypical femur fractures (AFF).
- He experienced a soft impact slip-and-fall which caused mid-shaft displaced fracture of the left femur diaphysis.
- Twelve weeks later he stepped oddly while walking and felt a pain in his thigh which then caused a fall; this proved to be a non-traumatic, mid-shaft non-displaced right femur fracture.
- He had no prior history of fractures, osteoporosis, bisphosphonates use, or any predisposing factors for bone disease.
- On review of laboratory results, it was noted that he had decreased levels of ALP for at least 9 years. After the two incidents of fracture, his ALP was found to be very low-normal.

## Laboratory Values and Imaging Studies



**Table 1:** Laboratory values for alkaline phosphatase over time, before and after fractures.

## Discussion

- Review of the literature suggests that initial presenting symptoms of adult HPP are usually bone pain related to pseudo-fractures or premature loss of teeth.
- Episodes of full thickness fracture of the femur diaphysis are reported in the setting of previously undiagnosed HPP and concurrent bisphosphonate therapy, which is known independent risk factor for AFF.
- This is the first case of bilateral AFF as the only presenting manifestation of adult HPP in an otherwise healthy individual.

## References

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**Figure 1:** Left Femur CT 3D Reconstruction Sept 2020



**Figure 2:** Right Femur XR Jan 2020