Histological Outcome of Hypophosphotaemic Rickets Treated with Phosphate Supplementation and Calcitriol for 6 years: A Case Report

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INTRODUCTION:

Hypophosphatemic rickets, a syndrome of low phosphate level with bone deformities and short stature has an estimated prevalence of 4.8 per 100,000. Long bones formed by process of enchondral ossification with provisional mineralization in cartilaginous epiphyseal plate, which then resorbed and replaced by osteoid matrix with mineralization. Morphologically there are enlargement and lateral expansion of osteochondral junction from the disrupted replacement of cartilage by osteoid matrix. Microfractures and stresses to the inadequately mineralized, weak, and poorly formed bone in paediatric patients resulted in bowing deformity. Histologically, unmineralized osteoid seen as a thickened layer of matrix. Treatment is aimed to correct hypophosphatemia and allow optimum bone growth by phosphate supplement and calcitriol.

REPORT:

A 15-year-old girl was referred to orthopaedic for bilateral lower limbs deformity secondary to hypophosphatemic rickets. She presented with progressive deformity over bilateral lower limbs at the age of 9. On examination, child's height was below 3rd centile, with bowing of bilateral femur and tibia. Widening and fraying of bilateral proximal tibia metaphysis with bowing seen on radiographs. Investigations revealed reduced serum phosphate with increased ALP, normal serum Vitamin D. She was given phosphate supplementation and calcitriol for past 6 years. Serial monitoring showed improved level of ALP and phosphate. No progression of bowing on radiographs after treatment. Open valgus corrective osteotomy for bilateral tibia and fibula, with plating of bilateral tibia was done at the age of 15. Intraoperatively, bone section from right proximal tibia was taken for HPE.

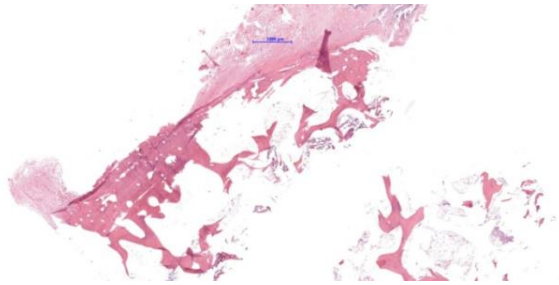

Figure 1: Irregular thick and thin bone trabecular, consistent with changes of Rickets

CONCLUSION:

Despite appropriate treatment with normalization of laboratory results, this patient's skeletal histology retained features of rickets.

REFERENCES:

1.McCarthy et al., The histology of metabolic bone disease. Diagnostic Histopathology (2016).
2. Marie-Eve Robinson et al., Mineralized tissues in hypophosphatemic rickets. Pediatric Nephrology. 2019