

Pathological Fracture Of Bilateral Neck Of Femur With Underlying Fanconi Syndrome : Red Flags In Management: A Case Report

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INTRODUCTION:

Fanconi syndrome is rare clinical entity with potentially devastating effects on bone mineral density. We present this case for its rarity and challenges involved in making a diagnosis.

MATERIALS & METHODS:

We report a case of 25-year- old lady who presented with bilateral lower limb weakness, bilateral hip pain and inability to walk. The weakness was present for 3 months, insidious in onset and progressively worsening. The onset of pain has a similar temporal origin as the weakness. Pain was aggravated by attempts to stand or walk. There was no preceding history of fall or trauma. There was also no bowel or urinary incontinence

On examination, patient is small statured with a weight was 30kg and height of 145cm. Neurologically, bilateral lower limbs were weak with motor power of L2 : 3/5, L3 : 3/5, L4 : 4/5, L5 : 4/5, S1 : 4/5. Sensory component was intact but deep tendon reflexes were diminished. There was no hip tenderness and passive movement of her hip joints elicited no pain.

Blood investigations revealed a normal full blood count, low levels of phosphate, calcium, potassium and bicarbonate with metabolic acidosis and high levels of alkaline phosphatase.

Serum PTH was raised with reduced 1,25 dihydroxycholecalciferol levels. Urinalysis demonstrated aciduria, glycosuria and proteinuria. Pelvic and lower limb radiographs revealed malunited bilateral neck of femur fracture with osteopenic bone quality.

RESULTS:

A diagnosis of Proximal renal tubular acidosis with Fanconi Syndrome was made. Osteomalacia was attributed to the Fanconi syndrome. Patient was started on phosphate and potassium supplements as well as Calcitriol. Her fractures were treated conservatively in view of had united. After 3 months, her electrolytes had returned to the normal range, she has no more weakness, is

able to ambulate and her fracture shows consolidation.

Figure 1: Pathological Fracture of Bilateral Neck of Femur



DISCUSSIONS:

Fanconi is a rare syndrome that consists of multiple defects in renal proximal tubular reabsorption. The condition is often marked by osteomalacia, acidosis and hypokalemia.¹ Osteomalacia associated with Fanconi's syndrome appears to respond well to calcium, phosphate, and vitamin D replacement.²

CONCLUSION:

Osteomalacia secondary to Fanconi syndromes offers a diagnostic challenge. A high index of suspicion could hasten the diagnosis and shorten the delay in initiating appropriate treatment.

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