A Girl with a Hard Painful Swelling at the Right Hip : Hyperphosphatemic Tumeral Calcinosis (mutation in the FGF23 gene)

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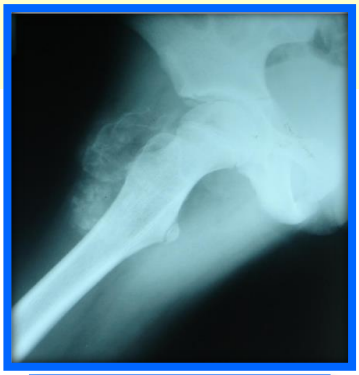
Introduction

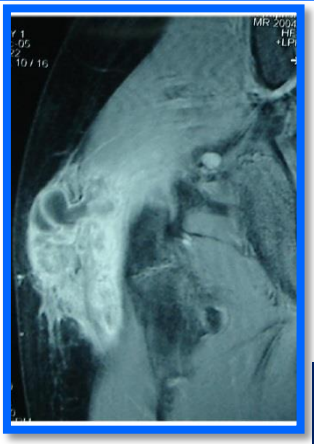
Hyperphosphatemic familial tumoral calcinosis (HFTC) is a condition characterized by an increase in the levels of phosphate in the blood (hyperphosphatemia) and abnormal deposits of phosphate and calcium (calcinosis) in in the soft tissue in the periarticular location outside the joint capsule

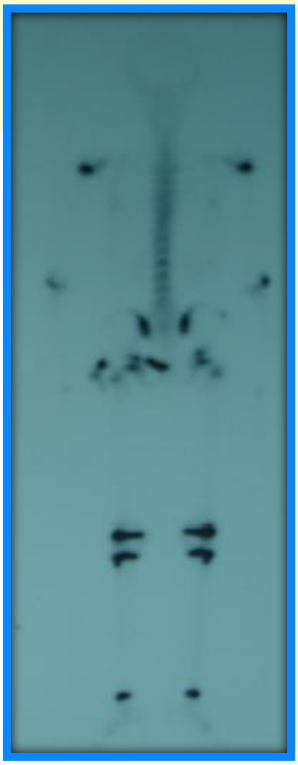
Case Report

This girl presented at the age of 10 years with swelling and pain in the right hip region. On examination a 10x10cm hard, non-tender swelling with well-defined margin was felt in upper lateral part of her left thigh.

Imaging the right hip revealed irregular calcified mass around the right hip mainly along the lateral aspect extending up to the upper part of the metaphysis arising mainly from the gluteus maximus extending laterally to the subcutaneous fat. Tc-99 MDP bone scintigraphy demonstrated abnormal uptake in the right gluteal soft tissue region.



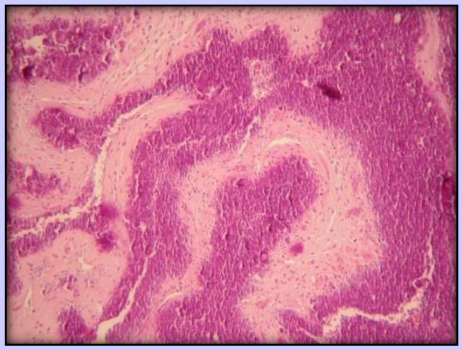




Marked soft tissue calcifications with soft tissue swelling at the lateral aspect of the upper thigh and hip region (plain x ray, MRI and bone scan

Results

Investigations showed high phosphorus, normal calcium, Alkaline phosphatase (ALP), and PTH, low 25(OH) cholecalciferol. Renal function was normal. The calcified mass was totally excised and histopathology and mutations in the FGF23 (chromosome 12) gene proved the diagnosis of HFTC.



Characteristic mixture of calcified material and giant cells (Hematoxylin &Eosin *x400*).

Treatment

Because of elevated serum phosphate she was started on phosphate binder (sevalamer 800mg 5 times daily) and nasal calcitonin twice daily in addition to dietary phosphate restriction.

On this treatment for seven years , serum phosphate ranged between 1.78 and 2.1mmol/L with normal serum calcium and ALP concentrations.

Clinical and MRI follow-up showed no evidence of recurrence and no renal or eye abnormalities nor hyperostosis for these 7 years. Linear growth was normal, (Ht SDS = 1.8, BMI = 16.5). She had normal pubertal development and menstruation.

Conclusion

10 years after excision the patient had no evidence of recurrence. She responded well to treatment with a phosphorus binder (sevalamer) and calcitonin as adjuvant therapy to surgery