Familial tumoral calcinosis caused by a novel FGF23 mutation: Response to induction of tubular renal acidosis with acetazolamide and the non-calcium phosphate binder sevelamer

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Hyperphosphatemic familial tumoral calcinosis (HFTC) is an uncommon disease characterized by periarticular calcifications produced by the deposition of amorphous extraosseous calcifications of hydroxyapatite. It is associated with hyperphosphatemia due to increased tubular phosphate reabsorption, despite normal renal function and normal plasma PTH levels. The disease can be caused by inactivating mutations in either the fibroblast growth factor 23 (FGF23) gene, the UDP-N-acetyl-D-galactosamine:polypeptide N-acetylgalactosaminyltransferase 3 (GALNT3) gene or in human KLOTHO (KL) gene. Herein, we describe a Caucasian 3-year-old girl with tumoral calcinosis who presented with elevated serum phosphorus levels and a large calcified mass at her left elbow which led to ulceration of the skin. Treatment with the phosphate binder sevelamer and the carbonic anhydrase inhibitor acetazolamide successfully reduced the serum phosphate levels and led to a reduction of the calcified mass. This medical