Rickets, elevated fibroblast growth factor-23 and mild anemia: Questions

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Case summary

A 4-year-old boy was referred to us for evaluation of wind-swept deformity of his lower extremities and X-ray finding of active rickets. Upon evaluation, we found that the deformities were noticed around age 2.5 years and since then had worsened. The child had otherwise normal history regarding his growth, development, and health. He took regular nutrition with appropriate intake of calcium and phosphorous, and no supplementations. He did not suffer from any chronic disease or took chronic medications. Both his parents and older sister were healthy and of normal height. On physical examination, we found him to be short statured (Z-score -1.95) with mild waddling gait, varus configuration of the right and valgus of the left leg, and widening of the wrist and ankle joints with no tenderness. Blood work showed creatinine 0.25 mg/dl (normal 0.26–0.64), calcium 9.7 mg/dl (normal 8.6–10.5), phosphorous 1.7 mg/dl (normal 3.5–6.8), 25(OH)-vitamin D 31 ng/ml (normal 30–50), 1,25(OH)₂-vitamin D 14 pg/ml (normal 18–64), parathyroid hormone (PTH) 57 pg/ml (normal 10–89), alkaline phosphatase (ALP) 887 IU/l (normal 10–320), and fibroblast-growth factor-23 (FGF-23) 806 RU/ml (normal < 230). Liver function tests were normal. The child's

hemoglobin was 11.5 g/dl (normal 11.5–13.5). Simultaneous random urine chemistries showed low tubular threshold for phosphate (TP/GFR) at 1.30 mg/dl and normal urinary calcium/creatinine ratio at 0.01 mg/mgCr. X-linked dominant hypophosphatemic rickets were suspected but the genetic test did not reveal a mutation in the *PHEX* gene.

Questions

1. What is the diagnosis?
2. What additional test(s), if any, would you obtain to confirm the diagnosis?
3. What would be the appropriate management of the child?

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