An unusual cause of severe rickets: Questions

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Case summary

A two-and-half-year-old girl presented with a history of failure to thrive, increasing abdominal distension, bowing of legs, and excessive tiredness for the past 1 year. There was history of polyuria and polydipsia since birth. She had a history of fracture of the left femur following trivial trauma 6 months previously, following which she was unable to walk. Her parents were non-consanguineous but there was a history of one male sibling death at 4 years of age with high-grade fever. He was also reported to have severe bowing of legs, pallor, and weakness.

Upon examination, the child's weight was 8.2 kg and height was 70 cm (both < third centile for age). She had clinical features of rickets, including a wide open anterior fontanel, widened wrists, rachitic rosary, and genu valgum. There was massive hepatosplenomegaly; liver 8 cm below costal margin, and spleen 6 cm below costal margin with ascites. Her eye examination was normal. Lab investigations revealed normal vitamin D levels (28 ng/ml), elevated serum alkaline phosphatase (2,458 IU/l), normal anion gap metabolic acidosis (pH 7.01, serum sodium 133 mmol/l, serum potassium 3.8 mmol/l, bicarbonate 17 mmol/l, chloride 106 mmol/l, anion gap-10) and severe hypophosphatemia (serum phosphate 0.7 mg%). Urine analysis revealed glucosuria, aminoaciduria, phosphaturia (elevated fractional excretion of phosphate: 63 %), and hypercalciuria. She had low platelet count (60,000/mm³) and deranged clotting studies (prothrombin time:

17.7 s; INR: 1.62; activated partial thromboplastin time 40.9 s). The rest of the liver function was within normal limits. X-ray of the lower limbs showed pathological fracture of left femur shaft and severe osteopenia (Fig. 1). Ultrasound of abdomen showed hepatosplenomegaly with coarse echo-texture and multiple hypodense lesions in the liver. The kidneys were normal-sized and there was no nephrocalcinosis.

Questions

1. What is the likely diagnosis in this child?
2. How will you confirm your diagnosis?
3. What are the treatment options?

Fig. 1 X-ray of femoral bones showing severe osteopenia and pathological fracture of shaft of left femur

The answers to these questions can be found at <http://dx.doi.org/10.1007/s00467-013-2522-8>.

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