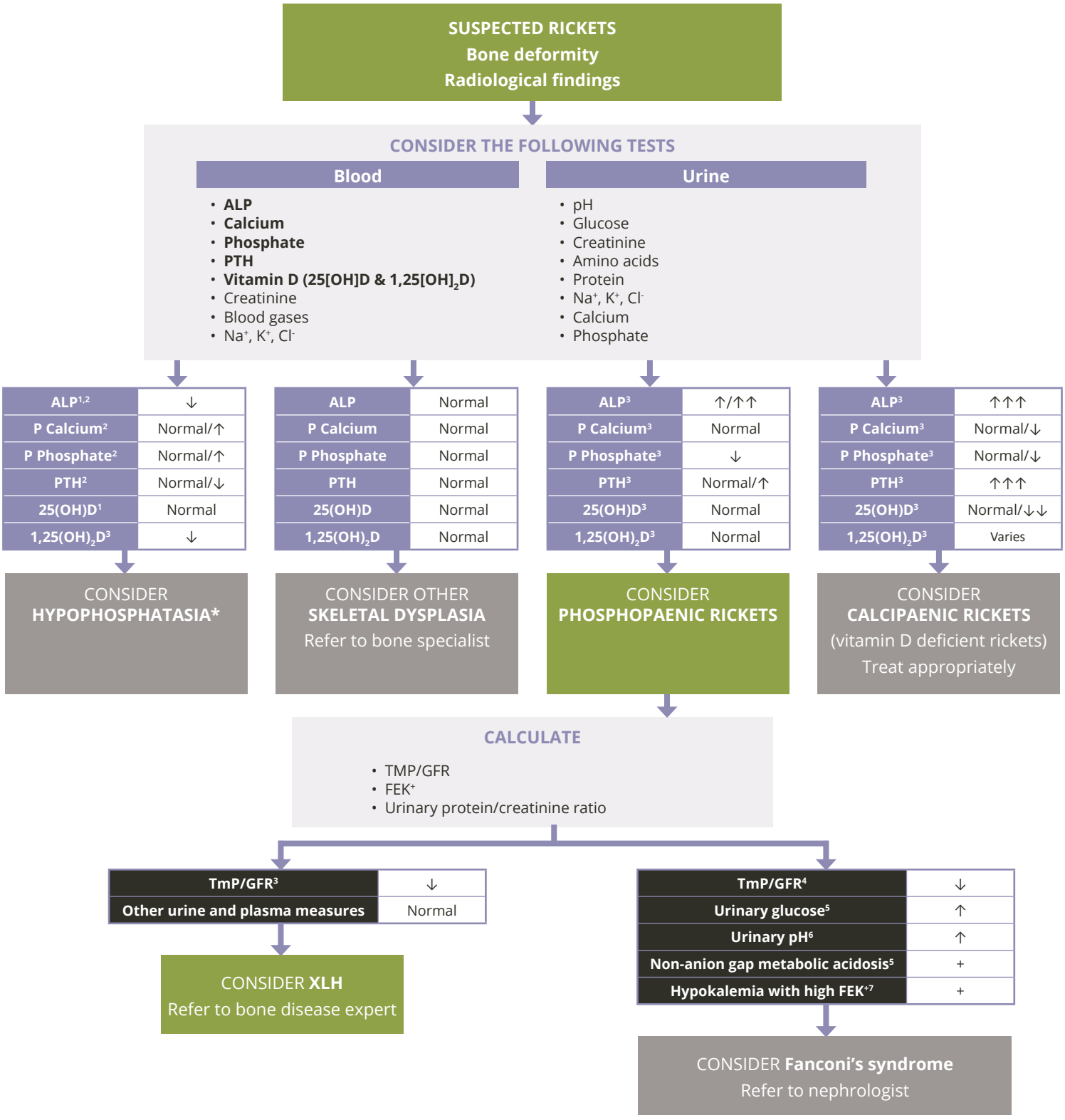
ASSESSMENT ALGORITHM FOR PAEDIATRIC PATIENTS WITH SUSPECTED X-LINKED HYPOPHOSPHATAEMIA (XLH)

This algorithm has been developed in collaboration with experts from the XLH Link Working Group.



↑, elevated; ↓, low; 1,25(OH)2D, 1,25-dihydroxyvitamin D (calcitriol); 25(OH)D, 25-hydroxyvitamin D (calcidiol); ALP, alkaline phosphatase; PTH, parathyroid hormone; TmP/GFR, ratio of tubular maximum reabsorption of phosphate to glomerular filtration rate; XLH, X-linked hypophosphataemia; P, plasma; +, present: FEK+, fractional excretion of potassium

*Measurement of serum vitamin B6, urinary phosphoethalonamine (PEA), inorganic pyrophosphate (PPI) and genetic testing of the *ALPL* gene are required for a definitive diagnosis of hypophosphatasia.

References

1. Simon, S et al. *Current Rheumatology Reports*. 2018; 20 (69):68-69; **2.** Linglart, A and Biosse-Duplan, *M. Curr Osteoporos Rep*. 2016;14:95–105; **3.** Haffner, D et al. *Nat Rev Nephrol*. 2019;15(7):435-455; **4.** *Payne, R B. Ann Clin Biochem*. 1998;35;201-206; **5.** Keefe P, Bokhari SRA. Fanconi Syndrome. [Updated 2018 Nov 28]. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2019 Jan-.Available from: https://www.ncbi.nlm.nih.gov/books/NBK534872/; **6.** Debray, F-G et al. *AJKD*. 2008; 51 (4): 691-696; **7.** Mustaqeem R, Arif A. Renal Tubular Acidosis. [Updated 2019 Jan 17]. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2019 Jan-. Available from: https://www.ncbi.nlm.nih.gov/books/NBK519044/

KKAP_CRV_01_OCT2020