

Images in pediatric endocrinology: vitamin D deficiency rickets and other nutritional deficiencies in a 12-month-old infant

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A girl of Indian ethnicity, born in Australia at term, presented at 12 months of age with irritability, failure to thrive, reluctance to weight bear and hypocalcemia. Her weight had decreased from the 25th to the 3rd percentile over 7 months. Her dietary intake had been predominantly breast and cow's milk. Clinically, she had tender, widened wrists (Figure 1). Blood tests were consistent with severe vitamin D deficiency: corrected calcium, 1.72 mmol/L (normal range 2.25–2.75 mmol/L); phosphate, 1.4 mmol/L (1.2–2.4 mmol/L); alkaline phosphatase, 1094 U/L (60–300 U/L); 25-hydroxy vitamin D, 11.2 nmol/L (50–150 nmol/L); and parathyroid hormone, 108 pmol/L (1–7 pmol/L). She had concomitant iron and vitamin B12 deficiencies: ferritin, 22 µg/L (30–150 µg/L) and vitamin B12 10 pmol/L (>35 pmol/L). The radiological appearance of her wrists and knees was consistent with rickets (Figure 2).



Figure 1 Clinical photograph displaying widened wrist.

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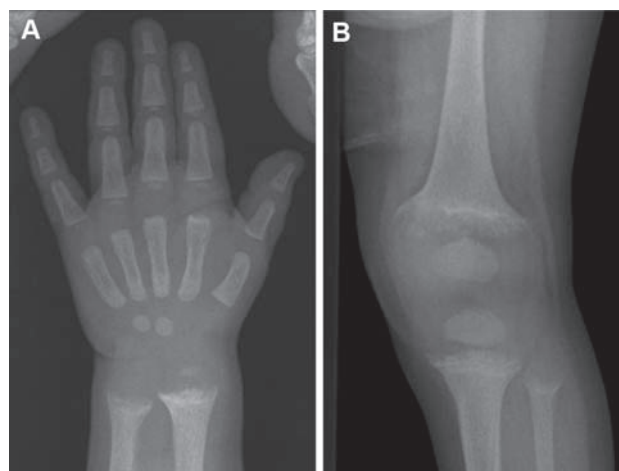


Figure 2 (A) Radiograph of the wrist showing metaphyseal irregularity with cupping and splaying and associated widening of the physes of the distal radius and ulna. (B) Radiograph of the knee showing similar changes in the distal femur and proximal tibia and fibula.



Figure 3 Radiograph of the wrist post-treatment showing resolution of the previously noted changes, with normal metaphyses and physes evident.

The child's mother had vitamin D and iron deficiencies, with a history of poor calcium intake: 25-hydroxy vitamin D, 21 nmol/L (50–150 nmol/L) and ferritin 9 µg/L (10–130 µg/L).

The patient was treated with calcium, vitamin D (initially calcitriol and later ergocalciferol), iron and vitamin B12. The quality and variety of her diet were also optimized. Within 6 months her clinical, biochemical and radiological features (Figure 3) were approaching normal. She had rapid progression in gross motor development. Her previous reluctance

to weight bear was probably due to joint pain and myopathy secondary to vitamin D deficiency.

Given her improvement with supplementation, this girl's presentation is consistent with nutritional vitamin D deficiency, with the additional contributions of dark skin color and maternal vitamin D deficiency. This case highlights the importance of investigating broadly for nutritional deficiencies, both in the infant and breastfeeding mother, when one deficiency is identified.