Primary Hyperparathyroidism: A Rare Cause of Genu Valgus in Adolescence

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A 15-year-old girl presented with a history of progressive genu valgus of both lower limbs. She had no other skeletal deformities or fractures. There was no significant family history, in particular none to suggest multiple endocrine neoplasia syndrome. Radiographs of the lower limbs confirmed severe bilateral genu valgus (Figure 1). Investigations revealed a markedly elevated serum corrected calcium of 12.4 mg/dL (normal, 8.8–10.4), phosphate of 2.8 mg/dL (normal, 3.4–5.8), alkaline phosphatase of 1136 IU/L (normal, 50–136), intact PTH of 1649 pg/ml (normal, 10–68), 25-hydroxyvitamin D of 28 ng/ml (normal, 9–52), and 24-hour urinary calcium of 196 mg/d (normal, 88–300). Further radiographs of the hand revealed classical changes of primary hyperparathyroidism: generalized osteopenia, subperiosteal resorption of the radial aspect of the middle phalanges (indicated by thick arrows) and terminal resorption with distal tufts (indicated by thin arrows).

Sporadic primary hyperparathyroidism (PHPT) in young patients is uncommon and is often associated with severe symptomatic end-organ damage compared to adults (1, 2). Bone disease occurred in one-third of young patients with PHPT, the second most common manifestation after renal stones (1–3). To date, there are only 3 reported cases of genu valgus as a manifestation of PHPT (3–5). Although most...
skeletal lesions resolve with parathyroidectomy, genu valgus often requires corrective surgery (3–5). A serum calcium level is a simple test to identify PHPT in the adolescent presenting with renal stones, bone disease, or pancreatitis.

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References