A severe presentation of vitamin D-dependent hypocalcemic rickets associated with hypophosphatemia

Uma apresentação grave de raquitismo hipocalcêmico dependente de vitamina D associado à hipofosfatemia

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This is a case report of a seven-year-old girl evacuated from Sao Tome and Principe due to rickets. She started walking at age 3, was unable to walk since age 4, and had recurrent episodes of respiratory distress[1]. At physical examination, she was malnourished, had bell-shaped thorax, deformed and varus limbs, and missing teeth. Blood tests revealed severe hypocalcemia (6.4 mg/dL) and hypophosphatemia (2.5 mg/dL), high alkaline phosphatase (4161 U/L) and

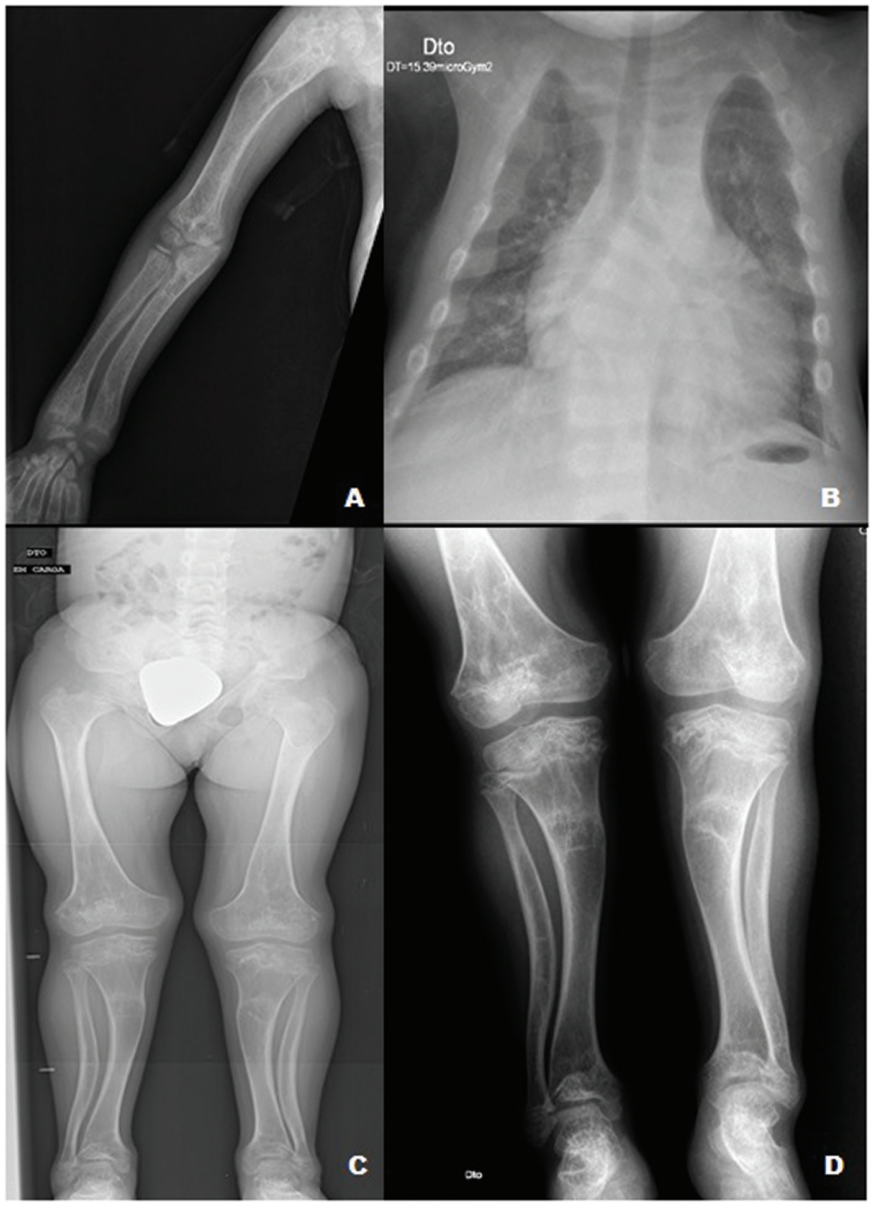

Figure 1. Patient radiographies showing severe bone deformities, pseudo-fractures in upper and lower limbs, and a bell-shaped thorax.

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parathyroid hormone (509.6 pg/ml), normal range 25-hydroxyvitamin D; 1,25-dihydroxyvitamin D was not performed. Genetic study identified mutations in both CYP27B1[2] gene copies, leading to alterations in alpha-1-hydroxylase function. A diagnosis of vitamin-D-dependent rickets type 1A was made.

Authors' Contribuitions

GV: conceptualization, investigation, methodology, writing of the original draft. LC: manuscript review and editing. TF: manuscript review and editing, formal analysis, supervision.

Conflict of Interest

The authors declare that they have no conflict of interest related to the publication of this manuscript.

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