Hypophosphatemic Rickets: Two unrelated Mexican female cases and review of the literature.


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Introduction
Hypophosphatemic Rickets (HR) is a rare genetic disorder characterized by poor growth, short disproportionate stature and lower limb anomalies. Clinical manifestations usually appear before one year of age, this entity is characterized by osseus hypomineralization, secondary to the increased expression of phosphonatins, especially FGF23, which acts in the sodium and phosphate co-transporters of the proximal convoluted tubule, inducing phosphaturia. HR exhibits AD, AR or X-linked inheritance patterns.

Case reports

Case 1
7.6-year-old girl, physical examination showed height 113 cm, weight 24 kg, arm span 116 cm, OFC 48 cm, with disproportionate short stature, genu varus, gait difficulties, asymmetric thorax, actually presenting normocalcemic, normophosphatemia, phosphaturia, calcitriol deficiency and secondary hyperparathyroidism.

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<th>Recent lab and ultrasound results</th>
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Kidney ultrasound
Kiddys with normal structures and parenchyma

Case 2
15-year-old girl at physical examination showed height 141 cm, weight 76 kg, with surgical history, the first one in order to correct bilateral tibia deformity at age 3, and genu varus correction surgery at age 5; at the moment the patient presents recidivant genu varo, normocalcemic, hypophosphatemia, and phosphaturia.

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Kidney ultrasound
Bilateral kidney microthiasis

Thyroid Gamma-gran
Parathyroid hyperplasia

Objective
We present two unrelated HR case reports, with clinical radiological and laboratory studies of Hypophosphatemic Rickets with osseous complications and adverse medical events.

Discussion
Hypophosphatemic rickets should be suspected in any child with disproportionate short stature, developmental delay and short, malformed legs. Only one third are due to nutritional deficit, clinical cases presented meet clinical criteria and radiological rickets. Case 1 presents normocalcemia and normophosphatemia indicating good control in the treatment. Case 2 has normocalcemia, hypophosphatemia, vitamin D insufficiency, bilateral kydey microthiasis and secondary hyperparathyroidism, which suggests a more aggressive disease presentation.

Conclusions
Due to functional complications in HR, early diagnosis and treatment is necessary, based in growth curves as well as serum values of phosphorus, calcitriol, alkaline phosphatase, and others, in order to offer an optimal medical care and prevent secondary osseous deformities.

REFERENCES