"Calcium, vitamin D and bone"

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

Nory Omayra Davalos, [1,2] Ruth Minerva Martínez, [3] Jose de Jesús Magallanes-Ordoñez,[4] Celia Andrea Ramírez-Aréchiga,[5] Enrique Cervantes-Pérez, [6] J.F. Michel Monrroy, [7] J. Fonseca Cárdenas, [7]

[1] Instituto de Genética Humana Dr. Enrique Corona, Doctorado de Genética Humana, CUCS, Universidad de Guadalajara, Guadalajara, Jalisoc, Mexico.
[2] Servicio de Genética, HRVGF ISSSTE, Zapopan, Jalisco, Mexico.
[3] Endocrinología, HRVGF ISSSTE, Zapopan, Jalisco, Mexico.
[4] Cirugía Plástica y Reconstructuvia, HRVGF ISSSTE, Zapopan, Jalisco, Mexico.
[5] CORENTE, Instituto Especializado en la CorInteligencia, S.C. D, Zapopan, Jalisco, Mexico.
[6] Departamento de Bienestar y Desarrollo Sustentable, Centro Universitario del Norte, Universidad de Guadalajara, Guadalajara, Jalisco, Mexico.
[7] Ortopedia y traumataología, HRVGF ISSSTE, Zapopan, Jalisco, Mexico.

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 phosphatonins , 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 spam 116 cm, OFC 48 cm, with disproportional short stature, *genu varo*, gait difficulties, asymmetric thorax; actually presenting normocalcemic, normophosphatemia, phosphaturia, calcitriol deficiency and secondary hyperparathyroidism.

Recent lab and ultrasound results

Serum levels	Calcium	9.6 mg/dl	Phosphorus	2.8 mg/dl	Creatinine	0.4 mg/dl	Vitamin D	5.08 ng/ml	PTH	82.8 pg/ml
Urine levels	Calcium	8.6 mg/dl	Phosphorus	9 mg/dl	Creatinine	24 mg/dl	Creatinine clearance		62.92 ml/min	
Kidney ultrasound	Kidneys 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 varum correction surgery at age 5; at the moment the patient presents recidivant *genu varo*, normocalcemic, hypophosphatemia, and phosphaturia.

Recent lab and imaging results

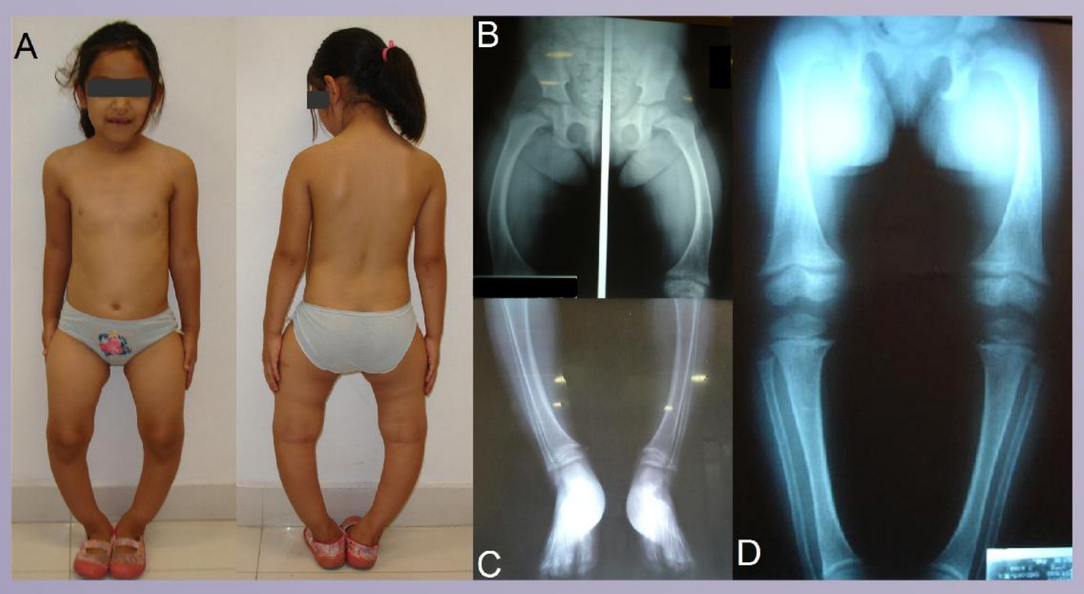
Serum levels	Calcium	9.5 mg/dl	Phosphorus	1.9 mg/dl	Creatinine	0.6 mg/dl	Vitamin D	13.8 ng/ml	PTH	78 pg/ml
Urine levels	Calcium	4.9 mg/dl	Phosphorus	8.2 mg/dl	Creatinine	39.9 mg/dl	Creatinine Clearance		64.65 ml/min	
Kidney ultrasound	Bilateral kydney microlithiasis									
Thyroid Gamma-gram	Parathyroid 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 kydney microlithiasis and secondary hyperparathyroidism, which suggests a more aggressive disease presentation.



A-C: Patient I, Important *genu varo* is appreciated. **B and C** Bilateral genu varum and adequate angle at the coxofemoral articulation is observed. **D: Patient II** Compatible changes with widening and arching of the metaphysis as well as reduction in the metaphyseal trabecular density, as well as bilateral Genu varum is noticed.

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.

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Davalos Nory
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