Cureus
Part of SPRINGER NATURE

Open Access Case Report

When Sunshine Is Not Enough: A Case Report of Vitamin D-Deficiency Rickets Causing Secondary Hyperparathyroidism in Florida

Review began 09/11/2026
Review ended 10/25/2026
Published 10/27/2026

© **Copyright** 2026
Colvin et al. This is an open access article distributed under the terms of the Creative Commons Attribution License CC-BY 4.0., which permits unrestricted use, distribution, and reproduction in any medium, provided the original author and source are credited.

DOI: 10.7759/cureus.118677

Kyle Colvin [1], Jorge Lima Gallardo [2], Ryan Chierico [3], Gabriella E. Kovalcik [4], Ernesto Tejeda Gomez [5], Jossias Genao-Cruz [5], Judith Cornely [5]

1. Orthopedic Surgery, Nova Southeastern University Dr. Kiran C. Patel College of Osteopathic Medicine, Fort Lauderdale, USA 2. Medicine, Nova Southeastern University Dr. Kiran C. Patel College of Osteopathic Medicine, Fort Lauderdale, USA 3. Osteopathic Medicine, Nova Southeastern University Dr. Kiran C. Patel College of Osteopathic Medicine, Fort Lauderdale, USA 4. Surgery, Nova Southeastern University Dr. Kiran C. Patel College of Osteopathic Medicine, Fort Lauderdale, USA 5. Pediatrics, Broward Health Medical Center, Fort Lauderdale, USA

Corresponding author: Kyle Colvin, kc2599@mynsu.nova.edu

Abstract

Rickets is a pediatric metabolic bone disorder caused by impaired growth plate mineralization, most commonly due to vitamin D deficiency. While classic manifestations include growth delay and bone deformities, acute presentations due to severe hypocalcemia are less common and may obscure the diagnosis. We describe a 13-month-old African-American girl who presented with bilateral hand stiffness and persistent thumb flexion secondary to profound hypocalcemia caused by severe vitamin D deficiency, highlighting an atypical neuromuscular presentation of nutritional rickets. The patient had no significant medical history and presented to the emergency department with a three-day history of subjective fever and a one-day history of bilateral finger stiffness and persistent thumb flexion. She was unable to walk and exhibited a downward trend in weight-for-age. Laboratory investigations revealed hypocalcemia (total calcium, 4.4 mg/dL; ionized calcium, 2.47 mg/dL), hypomagnesemia (1.3 mg/dL), elevated parathyroid hormone (210.1 pg/mL), and severe 25-hydroxyvitamin D deficiency (9.5 ng/mL). EKG demonstrated QT prolongation, and wrist radiographs showed metaphyseal fraying, widening, and cupping. Following calcium gluconate and magnesium infusions, the patient was admitted for further management of electrolyte abnormalities and underwent pediatric subspecialty evaluation for suspected nutritional rickets. This case highlights the diagnostic challenge of atypical nutritional rickets when acute metabolic derangements overshadow skeletal findings. Severe hypocalcemia can manifest as carpopedal spasm and cardiac conduction abnormalities, requiring prompt recognition. While inflammatory, infectious, and endocrine etiologies must be excluded, targeted laboratory and radiographic evaluation remains essential. Multidisciplinary management with calcium, vitamin D, and magnesium supplementation improved clinical and biochemical status. However, early readmission for recurrent hypocalcemia after initial discharge underscored the need for close outpatient follow-up. This case of vitamin D-deficiency rickets with secondary hyperparathyroidism and failure to thrive in South Florida underscores that adequate regional UV exposure does not preclude nutritional deficiency. Proper nutritional education, early recognition, and timely follow-up are essential for recovery and prevention of long-term sequelae. At approximately 14 months after presentation, she remained asymptomatic with a normalized 25-hydroxyvitamin D level of 44 ng/mL.

Categories: Nutrition, Pediatrics, Endocrinology/Diabetes/Metabolism
Keywords: carpopedal spasms, failure-to-thrive, hypocalcemia children, metaphyseal fraying, nutritional deficiency in infants, nutritional rickets, pediatric metabolic bone disease, qt-prolongation, secondary hyperparathyroidism, vitamin-d deficiency

Introduction

Rickets is a pediatric metabolic bone disorder characterized by impaired mineralization of the growing skeleton, most commonly due to deficiencies of vitamin D, calcium, or phosphate. This defect in endochondral ossification occurs at the growth plate, leading to skeletal weakness during periods of rapid growth. Vitamin D physiology underlies much of this disease process: cutaneous 7-dehydrocholesterol is converted to previtamin D3 on exposure to UV-B radiation, which is then hydroxylated in the liver to 25-hydroxyvitamin D (the primary circulating storage form and the analyte used to define deficiency) and subsequently in the kidney to the active hormone, 1,25-dihydroxyvitamin D (calcitriol). Deficiency is generally defined as a serum 25-hydroxyvitamin D level below 20 ng/mL, with levels of 20-29 ng/mL considered insufficient and levels below 10 ng/mL considered severely deficient. Although nutritional rickets is less common in developed countries than in many developing regions, it remains clinically significant, with an estimated annual incidence of 2.9-27 cases per 100,000 in the United States [1]. Although the incidence of nutritional rickets has declined in many developed countries, it remains a clinically significant and preventable condition, particularly among infants and young children with inadequate dietary intake, prolonged breastfeeding without sustained vitamin D supplementation, limited sunlight exposure, increased

skin pigmentation (which reduces cutaneous previtamin D3 synthesis for a given UV-B exposure), maternal vitamin D deficiency, or malabsorption syndromes [2].

Additionally, genetic forms of rickets include X-linked hypophosphatemic rickets (XLH) and vitamin D-dependent rickets (types I/II). XLH results from a mutation in the PHEX gene that increases FGF23 and impairs proximal renal tubular phosphate reabsorption [3]. In type I vitamin D-dependent rickets, a defect in the renal 1α-hydroxylase enzyme prevents conversion of vitamin D to its active form, calcitriol. In type II vitamin D-dependent rickets, end-organ resistance to vitamin D results from a mutation in the VDR gene [4]. The discussion further explores these genetic forms, focusing on how they were distinguished from nutritional rickets in this patient.

Rickets most often results from vitamin D, calcium, or phosphate deficiency. Vitamin D deficiency impairs intestinal absorption of calcium and phosphate. Low calcium levels increase parathyroid hormone (PTH) levels, leading to secondary hyperparathyroidism. Elevated PTH increases bone resorption, renal calcium reabsorption, and renal phosphate wasting, resulting in bone demineralization, normal-to-low-normal calcium, and hypophosphatemia in the early stages [5]. This decreases the calcium-phosphate product and impairs osteoid mineralization at the growth plates, causing bones to soften and growth plates to widen and become irregular [5].

Clinical manifestations of rickets vary with the severity and duration of mineral deficiency. Classic features include delayed growth, bowed lower extremities (genu varum or valgum), widened wrists, frontal bossing, and delayed fontanelle closure. Radiographic findings typically demonstrate widened and irregular growth plates with metaphyseal cupping and fraying [2]. In more severe or acute cases, metabolic abnormalities such as hypocalcemia, hypophosphatemia, hypomagnesemia, and secondary hyperparathyroidism may predominate, resulting in neuromuscular irritability, tetany, seizures, or cardiac conduction abnormalities, including QT interval prolongation [2].

Because acute metabolic manifestations may overshadow the underlying skeletal pathology, the diagnosis of rickets may be delayed if clinicians do not promptly consider it in infants presenting with neuromuscular or musculoskeletal symptoms. The literature has documented hypocalcemic tetany and carpopedal spasm as presenting features of previously undiagnosed rickets for decades, including a classic case of a toddler whose hand and foot stiffening prompted an emergency evaluation that ultimately revealed vitamin D-deficiency rickets [6]. More recently, atypical extraskeletal presentations of rickets driven by hypocalcemia, including gastrointestinal symptoms and other systemic complications, continue to be reported, reinforcing that neuromuscular or non-skeletal findings, rather than classic bony deformity, may be the presenting feature prompting diagnosis [7]. We present the case of a 13-month-old girl with severe vitamin D deficiency who presented with acute bilateral digital stiffness and persistent thumb flexion secondary to hypocalcemia. This case highlights an uncommon neuromuscular presentation of nutritional rickets and underscores the importance of early recognition and treatment to prevent serious complications.

Case Presentation

A 13-month-old African-American girl with no reported past medical history presented to the emergency department at Broward Health Medical Center with three days of subjective fevers and one day of bilateral finger stiffness and persistent thumb flexion. During physical examination, she cried whenever movement of her fingers was attempted. She had no gastrointestinal symptoms and was at baseline before admission. Pertinent developmental history included an inability to walk. She was born at 40 weeks and two days of gestation via spontaneous vaginal delivery and had perinatal hepatitis B exposure. Hepatitis B vaccine and hepatitis B immunoglobulin were administered. Maternal group B *Streptococcus* (GBS) was detected, and the mother received adequate intrapartum antibiotic prophylaxis. The patient was large for gestational age and did not require a NICU stay. Her nutritional history included breastfeeding until about two weeks before presentation at 13 months of age. Vitamin D supplementation was provided during the first few months of life but was not continued throughout breastfeeding. The patient was introduced to complementary foods (purees/solids) at six months of age. Maternal vitamin D status was not assessed, and previous pediatric follow-up records were unavailable. These limitations prevented a comprehensive assessment of the patient's nutritional and developmental history.

Vital signs were within normal limits except for an elevated blood pressure of 117/51 mmHg. This measurement was not repeated, and no additional renal imaging or renal-specific investigations were performed to evaluate the elevated blood pressure. Nephrology was consulted during hospitalization to manage calcium and vitamin D replacement rather than to evaluate hypertension. At presentation, weight was 8.8 kg (11.97th percentile), and length was 71.1 cm (6.87th percentile) on CDC growth standards. Physical examination revealed irritability, right tonsillar swelling, low-set ears, bilateral finger stiffness with persistent thumb flexion, a positive Trousseau sign (Figure *1*), and hyperpigmented macules on the back. Physical examination documented no classic rachitic skeletal findings (bowed legs, rachitic rosary, craniotabes, frontal bossing, or delayed fontanelle closure); the musculoskeletal findings supporting the diagnosis were radiographic, including metaphyseal fraying and cupping with physeal widening on left wrist radiography. No formal genetic testing was performed to evaluate the low-set ears or hyperpigmented macules. The growth chart (Figure *2*) shows a downward trend in weight-for-age. The mother also reported

food aversion and difficulties with breastfeeding and formula feeding.



FIGURE 1: Positive Trousseau sign

The image demonstrates the characteristic "main d'accoucheur" (obstetrician's hand) posture, with flexion of the wrist and metacarpophalangeal joints, extension of the interphalangeal joints, and adduction of the thumb across the palm.

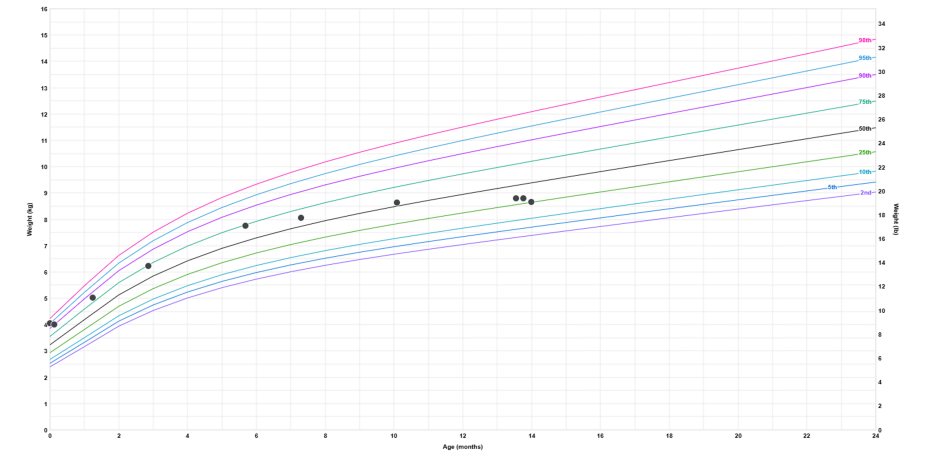


FIGURE 2: Weight-for-age percentile growth chart from the patient's electronic health record

Generated using Epic (Epic Systems Corporation, Verona, WI, USA).

Laboratory investigations were performed. A complete blood count showed a white blood cell count of 5.95 × 10^3/µL, slightly below the listed reference range, with otherwise unremarkable red cell indices and platelet count. A comprehensive metabolic panel revealed a bicarbonate level of 17 mEq/L, chloride level of 108 mEq/L, and calculated anion gap of 14. These findings raised the possibility of mild hyperchloremic metabolic acidosis, as discussed below; albumin was 4.3 g/dL, indicating that the patient's severe hypocalcemia was not an artifact of hypoalbuminemia (albumin-corrected calcium: approximately 4.2 mg/dL, essentially unchanged from the measured value). Additional laboratory findings showed hypocalcemia, hypomagnesemia, secondary hyperparathyroidism, severe 25-hydroxyvitamin D deficiency, hyperphosphatemia, and a markedly elevated alkaline phosphatase of 1,584 U/L (reference range 117-311

U/L), approximately five times the upper limit of normal, findings consistent with active rachitic bone disease (Table 1). Thyroid studies revealed a TSH level of 1.92 µIU/mL and a free T3 level of 3.29 pg/mL. Urine toxicology was obtained to evaluate for a toxic or ingestion-related cause of the patient's acute neuromuscular presentation and was negative. The patient also tested positive for COVID-19, which may account for the presenting fever; CRP and ESR were not obtained. The initial EKG showed normal sinus rhythm with a prolonged QT interval. A lateral radiograph of the left wrist (Figure 3) showed no fracture, dislocation, or mass. No significant soft tissue abnormalities were observed. Metaphyseal fraying and physeal widening were also evident. Metaphyseal cupping was also evident.

Parameter	Patient value	Reference range
Total calcium	4.4 mg/dL	8.8-10.8 mg/dL
Albumin-corrected calcium	~4.2 mg/dL	8.8-10.8 mg/dL
Ionized calcium	2.47 mg/dL	4.8-5.4 mg/dL (1.20-1.35 mmol/L)
Magnesium	1.3 mg/dL	1.6-2.6 mg/dL
Phosphorus	4.6 mg/dL (admission); peaked at 5.2 mg/dL during hospitalization	2.3-4.7 mg/dL
25-hydroxyvitamin D	9.5 ng/mL (checked once, at admission)	≥30 ng/mL (sufficient); <20 ng/mL (deficient); <10 ng/mL (severely deficient)
PTH (intact)	210.1 pg/mL	10-65 pg/mL
Sodium	139 mEq/L	135-145 mEq/L
Potassium	4.4 mEq/L	3.5-5.0 mEq/L
Chloride	108 mEq/L	98-108 mEq/L
Carbon dioxide (bicarbonate)	17 mEq/L	20-28 mEq/L
Anion gap	14	8-16
Blood urea nitrogen	3 mg/dL	5-18 mg/dL
Creatinine	0.3 mg/dL	0.2-0.4 mg/dL
Glucose	89 mg/dL	65-99 mg/dL
AST	64 U/L	0-53 U/L
ALT	13 U/L	<29 U/L
Alkaline phosphatase	1,584 U/L (first admission)	117-311 U/L
Total protein	6.7 g/dL	6.3-7.9 g/dL
Albumin	4.3 g/dL	4.4-5.4 g/dL
Total bilirubin	0.2 mg/dL	<1.2 mg/dL (term infant)
TSH	1.92 µIU/mL	~0.7-6.0 µIU/mL
Free T3	3.29 pg/mL	3.8-8.5 pg/mL
White blood cell count	5.95 x 10^3/µL	6.0-17.5 x 10^3/µL
Urine toxicology	Negative	Negative

TABLE 1: Admission laboratory values

PTH: parathyroid hormone, AST: aspartate aminotransferase, ALT: alanine aminotransferase, TSH: thyroid-stimulating hormone



FIGURE 3: Lateral radiograph of the left wrist demonstrating metaphyseal fraying and cupping with physeal widening

Based on the laboratory findings and radiographic evidence of rickets, the patient received multiple calcium gluconate infusions to correct hypocalcemia. The patient was admitted to the acute care unit for further management of hypocalcemia and hypomagnesemia, with subsequent monitoring of phosphorus levels, requiring evaluation by pediatric specialists for suspected rickets secondary to malnutrition. Laboratory evaluation during this first hospitalization showed decreased ionized calcium (3.29 mg/dL; total calcium 5.6 mg/dL) and magnesium (1.6 mg/dL), despite starting oral calcium supplementation, vitamin D (cholecalciferol 25 mcg (~1,000 IU)), and calcitriol 0.1 mcg daily. The attending physician increased the daily cholecalciferol dose from 1,000 IU (25 mcg) to 2,000 IU (50 mcg) and maintained it thereafter. The patient also received 220 mg of magnesium sulfate for hypomagnesemia, and magnesium improved to 1.9 mg/dL (reference range 1.7-2.3 mg/dL) 2 days before discharge. The medical team recommended continued hospitalization until total calcium exceeded 8.0 mg/dL and ionized calcium exceeded 4.0 mg/dL, with ionized calcium considered the preferred monitoring parameter [8]; however, the family requested discharge before these goals were met due to social circumstances, including a parental job loss and the need to care for another child at home. Serial EKG monitoring was performed given the initial QT prolongation, though a corrected QT (QTc) value and correction formula were not calculated or reported on the cardiology tracings at any point during admission: repeat tracings subsequently showed a nonspecific ST abnormality with persistent QT prolongation, then right axis deviation with a nonspecific T-wave abnormality, then right ventricular hypertrophy with a repolarization abnormality and possible biventricular hypertrophy, and finally resolved to a normal sinus rhythm with only nonspecific ST changes and baseline artifact; a formal echocardiogram performed during this monitoring showed normal segmental cardiac anatomy, suggesting

the interval EKG changes most likely reflected the normal right-heart-dominant axis and voltage pattern common in infants under one year of age rather than structural pathology. The patient was discharged with calcitriol 1 mcg/mL, 0.1 mL PO daily (0.1 mcg/day), calcium carbonate 1250 mg/5 mL, 1 mL PO Q6h (250 mg calcium carbonate per dose, ~100 mg elemental calcium per dose, ~400 mg/day elemental calcium at four doses daily, approximately 45.5 mg/kg/day based on the patient's 8.8 kg weight), and cholecalciferol 10 mcg/mL, 5 mL PO daily (50 mcg, ~2,000 IU/day) [9].

Two days after initial discharge, the patient returned and was readmitted to the pediatric acute care unit after a repeat basic metabolic panel (BMP) revealed recurrent hypocalcemia (total calcium, 6.4 mg/dL). Urine calcium was 1.1 mg/dL at initial presentation and 1.5 mg/dL five days later at first discharge, though no laboratory reference range was established for comparison. During the second hospitalization, which lasted three nights, calcium levels improved following treatment with calcitriol 0.1 mcg orally once daily, calcium carbonate 250 mg orally every six hours (~100 mg elemental calcium per dose, ~400 mg/day elemental calcium at four doses daily, approximately 45.5 mg/kg/day), cholecalciferol 50 mcg PO daily (~2,000 IU/day), and a high-calcium diet (Table *2*) [9]. The calcium carbonate regimen was then switched to 375 mg PO three times daily (~150 mg elemental calcium per dose, ~450 mg/day elemental calcium, approximately 51.1 mg/kg/day) per nephrology and endocrine recommendations, and this dose was continued through final discharge.

Hospitalization day	Total calcium (mg/dL)	Ionized calcium (mg/dL)	Alkaline phosphatase (U/L)
Readmission day 1	6.4	-	1749
Readmission day 1 (repeat)	7.5	-	-
Readmission day 3	7.9	-	-
Readmission day 4 (discharge)	8.8	4.77	1494

TABLE 2: Total calcium, ionized calcium, and alkaline phosphatase trend during the second hospitalization (readmission)

Reference ranges: total calcium 8.8-10.8 mg/dL; ionized calcium 4.8-5.4 mg/dL; alkaline phosphatase 117-311 U/L. Discharge criteria were total calcium >8.0 mg/dL and ionized calcium >4.0 mg/dL.

According to the primary care provider and the patient's mother, food aversion initially raised concern for avoidant/restrictive food intake disorder (ARFID), which can be associated with nutritional deficiencies and psychosocial impairment [10]; ARFID was considered as a differential concern but was not the gastroenterology team's formal diagnosis, which was food aversion. To address the food aversion, Pediatric Gastroenterology and Nutrition was consulted. Additional workup included a celiac panel, which was negative; fecal calprotectin, which was 63 µg/g (borderline elevated, given a laboratory-reported range of <50 normal, 60-120 borderline, and >120 elevated); and fecal pancreatic elastase, which was 125 µg/g, falling within the mild-to-moderate exocrine pancreatic insufficiency range (<100 severe insufficiency, 100-200 mild-to-moderate insufficiency, >200 normal, per the reporting laboratory). Dietary recommendations included a regular toddler diet high in calcium, with calorie goals of 898-1000 kcal/day, 12-14 grams of protein/day, and 800 mL/day of fluids, along with Pediasure three times daily. Intravenous access was maintained using a saline lock.

On the day of discharge, calcium levels reached 8.8 mg/dL and ionized calcium levels reached 4.77 mg/dL, meeting discharge criteria of total calcium above 8 mg/dL and ionized calcium above 4 mg/dL; alkaline phosphatase at discharge was 1494 U/L, improved from 1749 U/L earlier in the second admission but still markedly above the normal range, consistent with ongoing but improving osteoblastic activity during bone healing. The patient was cleared for discharge by pediatric specialists and instructed to continue medications at home and to monitor intake. The patient has since followed up with her primary care provider for over a year and has remained asymptomatic. Her 25-hydroxyvitamin D level was 20 ng/mL about five months after the index admission and normalized to 44 ng/mL at her most recent visit, about 14 months after the index admission. Weight and length at both hospitalizations are compared in Table *3*.

Measurement	First admission	Second admission (readmission)
Weight	8.8 kg (11.97th percentile)	8.8 kg (10.81st percentile)
Length	71.1 cm (6.87th percentile)	71.1 cm (5.77th percentile)
Head circumference	Not measured	Not measured

TABLE 3: Weight and length at first admission and readmission (CDC growth standards)

Discussion

Nutritional rickets is uncommon in South Florida, where sunlight is abundant throughout the year. Nevertheless, vitamin D-deficiency rickets can occur in children with inadequate nutrient intake or other risk factors and is more common in northern regions with reduced UV-B exposure [11]. Beyond latitude, skin pigmentation itself is a well-established biological risk factor: melanin competes with 7-dehydrocholesterol for UV-B photons in the epidermis, meaning that for a given sun exposure, more heavily pigmented skin synthesizes substantially less previtamin D3 than lighter skin [5]. This mechanism helps explain why an infant living in a sunny region such as South Florida may still develop severe nutritional rickets despite abundant sunlight exposure.

In this case, malnutrition was considered a probable contributor to the patient's rickets, given the downward trend in weight-for-age, prolonged breastfeeding without sustained vitamin D supplementation, and laboratory evidence of micronutrient deficiencies. Vitamin D deficiency commonly lowers serum calcium and triggers secondary hyperparathyroidism, contributing to nutritional rickets [12,13]. Physical examination findings such as carpopedal spasm, laboratory findings including calcium, phosphate, and PTH levels, and imaging findings such as metaphyseal cupping on wrist radiographs collectively support the diagnosis of rickets. Although the patient did not undergo genetic testing for XLH or vitamin D-dependent rickets, several clinical and biochemical findings favored a primarily nutritional etiology. XLH classically causes renal phosphate wasting and resultant hypophosphatemia through excess FGF23 activity, whereas this patient's phosphorus levels were normal to mildly elevated rather than low, a pattern inconsistent with unopposed phosphate wasting. Similarly, the patient's severely low 25-hydroxyvitamin D level and her clinical and biochemical response to standard cholecalciferol and calcium repletion are more consistent with nutritional vitamin D deficiency than with vitamin D-dependent rickets, in which standard cholecalciferol dosing is typically ineffective due to a defect in either activation of or response to vitamin D itself.

Several conditions in this patient's differential diagnosis highlight the diagnostic challenge of distinguishing inflammatory, infectious, and metabolic etiologies of musculoskeletal and neuromuscular symptoms in young children. Juvenile idiopathic arthritis and reactive arthritis may present with joint stiffness and limited range of motion; however, these diagnoses require excluding infectious causes such as septic arthritis and osteomyelitis, which often cause systemic toxicity and localized joint findings [14]. In contrast, metabolic disorders resulting in hypocalcemia, such as vitamin D deficiency, hypoparathyroidism, renal disease, and DiGeorge syndrome, can present with neuromuscular irritability, carpopedal spasm, and cardiac conduction abnormalities [2,15]. Careful interpretation of laboratory findings, including calcium, phosphate, magnesium, and PTH levels, along with characteristic radiographic findings such as metaphyseal cupping and fraying, is essential to distinguish nutritional rickets from these alternative diagnoses [2]. The patient's admission chemistry panel also revealed a bicarbonate level of 17 mEq/L, chloride level of 108 mEq/L, and calculated anion gap of 14. These findings raised the possibility of mild hyperchloremic, normal-anion-gap metabolic acidosis and prompted consideration of renal tubular acidosis (RTA) in the differential diagnosis, since proximal (type 2) RTA can itself cause phosphate wasting and rickets-like bone disease independent of vitamin D status. Urine calcium was 1.1 mg/dL on admission and 1.5 mg/dL at discharge from the first hospitalization, though no formal urine pH, fractional excretion of bicarbonate, renal ultrasound, or other dedicated RTA-specific testing was obtained at any point during either admission, so this differential cannot be formally excluded on the available workup; the patient's overall biochemical and radiographic picture was nonetheless most consistent with nutritional vitamin D deficiency rather than a primary renal tubular process. No genetic testing was performed to evaluate the patient's low-set ears or hyperpigmented macules, so a contributing or coincidental genetic syndrome, including 22q11.2 deletion syndrome, likewise cannot be formally excluded and is acknowledged as a limitation of this case.

In this case, reduced cutaneous vitamin D synthesis and inadequate nutritional intake were considered complementary contributing factors: the patient's skin pigmentation would be expected to lower previtamin D3 synthesis for any given sun exposure, while prolonged breastfeeding without sustained vitamin D supplementation removed the dietary buffer that would ordinarily offset that reduced cutaneous synthesis. Malnutrition was the most immediate modifiable contributor to the patient's rickets, supported by a documented downward trend in weight-for-age, food aversion with feeding difficulty, and breastfeeding continued nearly to presentation without sustained vitamin D supplementation beyond early infancy.

Gastroenterological investigations for possible malabsorption revealed a negative celiac panel, borderline-elevated fecal calprotectin, and a fecal pancreatic elastase level within the range associated with mild-to-moderate exocrine pancreatic insufficiency; while these findings do not establish a specific malabsorptive diagnosis and celiac disease was excluded, the elastase result raises the possibility of a co-existing, milder degree of malabsorption compounding the patient's primarily nutritional vitamin D and calcium deficiency and would merit outpatient follow-up and consideration of further pancreatic function evaluation if persistent.

Rickets is best diagnosed by combining compatible clinical, biochemical, and radiographic findings, including bone pain, deformities, and growth impairment, along with low serum 25-hydroxyvitamin D, hypocalcemia, elevated alkaline phosphatase, and hypophosphatemia [16]. Metaphyseal changes such as cupping, fraying, and widening of the long bones may be appreciated on radiography. Lastly, genetic and biochemical testing may distinguish nutritional from genetic rickets. Standard treatment consists of a dual approach of oral vitamin D and calcium supplementation. It is important to distinguish prevention from treatment dosing in this context: the Endocrine Society's 300-2,000 IU/day range is intended for the prevention of nutritional rickets in the general pediatric population, depending on age and weight [17]. By contrast, the 2016 Global Consensus Recommendations on Prevention and Management of Nutritional Rickets specify considerably higher treatment doses for children with established disease; for a child in this patient's age range (>12 months), 3,000-6,000 IU/day for 90 days, followed by a maintenance dose of 600 IU/day [18]. The patient's cholecalciferol dosage (approximately 1,000 IU/day initially, increased to approximately 2,000 IU/day at discharge and after readmission) was below the consensus-recommended treatment range for her age. This may have contributed to the early recurrence of hypocalcemia. This patient was treated with aggressive calcium repletion, including oral calcium carbonate and calcitriol, in conjunction with cholecalciferol supplementation and magnesium replacement for concurrent hypomagnesemia; calcitriol, an active vitamin D analog not typically used in isolated nutritional rickets, was used here adjunctively for rapid correction of severe, symptomatic hypocalcemia given the patient's cardiac and neuromuscular presentation, rather than as standard maintenance therapy. Calcitriol was continued at discharge from both hospitalizations; the planned total duration of calcitriol therapy was not specified in the available chart documentation, and its discontinuation was most likely intended to be guided by the patient's ongoing biochemical response (normalization of calcium and resolution of secondary hyperparathyroidism) rather than a fixed treatment length, consistent with typical adjunctive use in this setting, though this rationale was not explicitly documented. Serial electrolyte monitoring and EKG monitoring were performed due to initial QT prolongation. Following multidisciplinary management involving pediatric nephrology, gastroenterology, and nutrition services, the patient's calcium levels normalized and her neuromuscular symptoms resolved. She was subsequently discharged with recommendations for a high-calcium diet and continued supplementation.

The patient's readmission for recurrent hypocalcemia two days after initial discharge highlights the importance of ensuring biochemical stability before discharge. In this instance, the earlier discharge occurred before the treating team's preferred biochemical thresholds were met, at the family's request, due to social circumstances including a parental job loss and the need to care for another child at home, underscoring that psychosocial and logistical pressures on a family can directly shape the clinical course, independent of the underlying disease severity. Beyond these social circumstances, relying solely on a single calcium measurement exceeding a predefined threshold may be insufficient during ongoing skeletal remineralization: as PTH-driven bone resorption resolves and osteoblastic activity resumes, bone tissue can rapidly reincorporate circulating calcium and phosphate, a phenomenon commonly termed "hungry bone syndrome," transiently outpacing oral repletion and precipitating recurrent hypocalcemia even after apparent initial correction [8]. This case suggests clinicians should anticipate both possibilities: proactively addressing modifiable social barriers to safe discharge and maintaining a low threshold for close, short-interval outpatient follow-up in similarly severe presentations, particularly when treatment doses fall below consensus-recommended levels or when discharge occurs before treatment goals are fully met. This report is limited by the lack of a documented sun exposure history, repeat PTH measurements, follow-up radiographs documenting healing, and repeat 25-hydroxyvitamin D testing during hospitalization (outpatient values are described above). Recovery during the hospitalization was tracked primarily by serial total and ionized calcium and, during the second admission, alkaline phosphatase. Additionally, the patient’s pre-admission dietary intake was not quantified.

Conclusions

This case illustrates an atypical presentation of nutritional rickets characterized by acute neuromuscular symptoms secondary to severe hypocalcemia in a young child. Although rickets is often associated with classic skeletal deformities and delayed growth, this report underscores that metabolic complications such as hypocalcemia, along with associated cardiac and neuromuscular abnormalities, may be the initial and predominant clinical features. Recognition of these presentations is critical, as delayed diagnosis can lead to potentially life-threatening complications.

Early laboratory evaluation, radiographic imaging, and multidisciplinary management were essential to establish the diagnosis and correct the metabolic abnormalities in this patient. Prompt calcium, vitamin D, and magnesium supplementation led to clinical and biochemical improvement. This case highlights the

continued occurrence of nutritional rickets in developed countries and underscores the importance of considering this diagnosis in infants presenting with unexplained musculoskeletal rigidity or electrolyte abnormalities. Increased awareness and early intervention remain key to preventing morbidity associated with this largely preventable condition.

Additional Information

Author Contributions

All authors have reviewed the final version to be published and agreed to be accountable for all aspects of the work.

Concept and design: Kyle Colvin, Judith Cornely, Ernesto Tejeda Gomez, Jossias Genao-Cruz, Gabriella E. Kovalcik, Jorge Lima Gallardo, Ryan Chierico

Acquisition, analysis, or interpretation of data: Kyle Colvin, Judith Cornely, Ernesto Tejeda Gomez, Jossias Genao-Cruz, Gabriella E. Kovalcik, Jorge Lima Gallardo, Ryan Chierico

Drafting of the manuscript: Kyle Colvin, Judith Cornely, Gabriella E. Kovalcik, Jorge Lima Gallardo, Ryan Chierico

Critical review of the manuscript for important intellectual content: Kyle Colvin, Judith Cornely, Ernesto Tejeda Gomez, Jossias Genao-Cruz, Gabriella E. Kovalcik, Jorge Lima Gallardo, Ryan Chierico

Supervision: Judith Cornely, Ernesto Tejeda Gomez, Jossias Genao-Cruz

Disclosures

Human subjects: Informed consent for treatment and open access publication was obtained or waived by all participants in this study. **Conflicts of interest:** In compliance with the ICMJE uniform disclosure form, all authors declare the following: **Payment/services info:** All authors have declared that no financial support was received from any organization for the submitted work. **Financial relationships:** All authors have declared that they have no financial relationships at present or within the previous three years with any organizations that might have an interest in the submitted work. **Other relationships:** All authors have declared that there are no other relationships or activities that could appear to have influenced the submitted work.

References

1. Gentile C, Chiarelli F: Rickets in children: an update. Biomedicines. 2021, 9:738. 10.3390/biomedicines9070738
2. Dahash BA, Sankararaman S: Rickets. StatPearls [Internet]. StatPearls Publishing, Treasure Island (FL); 2023.
3. Carpenter TO, Imel EA, Holm IA, Jan de Beur SM, Insogna KL: A clinician's guide to X-linked hypophosphatemia. J Bone Miner Res. 2011, 26:1381-8. 10.1002/jbmr.340
4. Feldman D, J Malloy P: Mutations in the vitamin D receptor and hereditary vitamin D-resistant rickets . Bonekey Rep. 2014, 3:510. 10.1038/bonekey.2014.5
5. Holick MF: Resurrection of vitamin D deficiency and rickets . J Clin Invest. 2006, 116:2062-72. 10.1172/JCI29449
6. Duplechin RY, Nadkarni M, Schwartz RP: Hypocalcemic tetany in a toddler with undiagnosed rickets . Ann Emerg Med. 1999, 34:399-402. 10.1016/s0196-0644(99)70137-x
7. Francioni A, Simone V, Laschi E, et al.: Case report: atypical presentation of rickets with hypocalcemia-related emesis. Front Pediatr. 2025, 13:1627190. 10.3389/fped.2025.1627190
8. Roy A, Chowdhury AS, Ray A, et al.: Diagnostic approach to rickets: an Endocrine Society of Bengal (ESB) consensus statement. Ann Pediatr Endocrinol Metab. 2024, 29:284-307. 10.6065/apem.2448044.022
9. The Johns Hopkins Hospital, Fatola T, Heckman J, Silvestri NJ: The Harriet Lane Handbook: A Manual for Pediatric House Officers. Elsevier, Philadelphia (PA); 2026.
10. American Psychiatric Association: Diagnostic and Statistical Manual of Mental Disorders. American Psychiatric Publishing, Washington, D.C.; 2022. 10.1176/appi.books.9780890425787
11. Rajakumar K, Thomas SB: Reemerging nutritional rickets: a historical perspective . Arch Pediatr Adolesc Med. 2005, 159:335-41. 10.1001/archpedi.159.4.335
12. Narasimhan S, Lavik A, Auron M: Rickets. Pediatr Rev. 2025, 46:494-509. 10.1542/pir.2024-006494
13. Chanchlani R, Nemer P, Sinha R, et al.: An overview of rickets in children . Kidney Int Rep. 2020, 5:980-90. 10.1016/j.ekir.2020.03.025
14. Ravelli A, Martini A: Juvenile idiopathic arthritis. Lancet. 2007, 369:767-78. 10.1016/S0140-6736(07)60363-8
15. Cooper MS, Gittoes NJ: Diagnosis and management of hypocalcaemia . BMJ. 2008, 336:1298-302. 10.1136/bmj.39582.589433.BE
16. Carpenter TO, Shaw NJ, Portale AA, Ward LM, Abrams SA, Pettifor JM: Rickets. Nat Rev Dis Primers. 2017, 3:17101. 10.1038/nrdp.2017.101
17. Demay MB, Pittas AG, Bikle DD, et al.: Vitamin D for the prevention of disease: an Endocrine Society clinical

practice guideline. J Clin Endocrinol Metab. 2024, 109:1907-47. 10.1210/clinem/dgae290
18. Munns CF, Shaw N, Kiely M, et al.: Global consensus recommendations on prevention and management of nutritional rickets. J Clin Endocrinol Metab. 2016, 101:394-415. 10.1210/jc.2015-2175