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CASE REPORT

Pseudohypoparathyroidism Type 1a with Hypocalcemia in a 25-Year-Old Woman: A Case Report of Delayed Presentation

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Abstract

Pseudohypoparathyroidism (PHP) is a rare endocrine disorder caused by end-organ resistance to parathyroid hormone (PTH), resulting in hypocalcemia and hyperphosphatemia despite elevated PTH levels. PHP type 1a is associated with Albright hereditary osteodystrophy (AHO) features such as brachydactyly, short stature, and a round face. Usually these group of patients present in early childhood. In his case report, we found a 25-year-old female with fatigue and muscle cramps. She had short stature (101 cm), brachydactyly, a round face, and a positive knuckle dimple sign. Investigations revealed hypocalcemia (5.6 mg/dL), hyperphosphatemia (2 mmol/L), normal magnesium and albumin levels, and elevated PTH (70.1 pg/mL). Radiographs revealed shortened fourth and fifth metacarpals. Genetic and cAMP testing were unavailable. Treatment with oral calcium carbonate (1.5 g/day) and calcitriol (0.5 mcg/day) normalized calcium and resolved symptoms within one month. In low-resource settings, recognition of the AHO phenotype with basic biochemical evidence can enable timely PHP diagnosis. Early calcium and vitamin D therapy is essential to prevent complications.

Keywords: Pseudohypoparathyroidism, Albright hereditary osteodystrophy, Brachydactyly, Hyperphosphatemia, Elevated PTH, Hypocalcemia

Introduction

PHP is a heterogeneous disorder with prevalence of the disorder is about 0.79 per 100,000.[1] End-organ unresponsiveness to parathyroid hormone is a typical characteristic of this condition, which is caused by genetic abnormalities in the hormone receptor adenylate cyclase system. Parathyroid hormone (PTH) is a significant hormone which plays a role in the regulation of serum calcium. As the target organ is resistant to PTH it leads to a disrupted balance in calcium and phosphate homeostasis. PTH plays a role in the regulation of serum calcium.[2] Patients typically present with features of hypocalcemia, such as muscle cramps, seizures, or tetany, and biochemical findings include low serum calcium, high serum phosphate, and elevated PTH levels.

PHP-1 is characterized by resistance to parathyroid hormone (PTH) and Albright's hereditary osteodystrophy (AHO) phenotype, which includes brachydactyly, small stature, round face, subcutaneous ossifications, and early-onset obesity.[3] PHP Types 1A and 1C are both characterized by heterogeneous clinical findings, such as brachydactyly, rounded face, short stature, central obesity and subcutaneous calcification, and mental retardation.[4] Maintaining calcium levels is one of the main functions of parathyroid hormone. Parathyroid-related hypocalcemia and hyperphosphatemia may result from insufficient production of PTH by the parathyroid glands (hypoparathyroidism) or from resistance to its action on target organs (Pseudohypoparathyroidism).[5]

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Case presentation

A 25-year-old female came to our outpatient department with the complaints of fatigue and intermittent episodes of muscle cramping over several months. But she has no remarkable past medical history. On physical examination, she exhibited hallmark phenotypic features consistent with pseudohypoparathyroidism type 1a such as short stature and neck, round face, short stubby fingers

and brachydactyly as seen in Figure 1. She weighed 24 kg, measured 101 cm in height significantly below the fifth percentile for age. Her vital signs were within normal limits and all the systemic examinations were unremarkable. The knuckle dimple sign was positive in hands which also supported the clinical suspicion (figure-1). Chvostek's and Trousseau's signs were both negative indicating the absence of overt hypo calcemic neuromuscular irritability at the time of examination. She was a product of non-consanguineous marriage with a normal birth, but her developmental milestones were delayed which added weight to the diagnosis. Her family history was also non-significant. Initial laboratory investigation (table-I) results revealed hypocalcemia (5.6 mg/dL) with hyperphosphatemia (2 mmol/L) with normal albumin (4.3 g/dl) and magnesium (2 mg/dl) level. Later serum PTH level was also found high (70.1pg/ml). X-ray of hand shows shortness of fourth and fifth metacarpal bones (figure 2). Thyroid function test and other endocrine test results were normal as shown in Table 1. PTH induced cAMP levels and genetic analysis could not be done due to non-availability of the tests in the country.

The diagnosis of Pseudohypoparathyroidism type 1a was made based on clinical examination and the biochemical profile showing hypocalcemia, hyperphosphatemia, and elevated PTH levels, along with phenotypic features suggestive of Albright hereditary osteodystrophy (AHO). She was treated with regular oral calcium carbonate (1.5 gm daily) and oral calcitriol (0.5 mcg/day). Her symptoms improved significantly with the treatment. After 1 month of follow-up, the patient was completely asymptomatic. Her follow-up serum corrected calcium level was normal and PTH was high normal (60 pg/ml).

Table 1: Comprehensive overview of all relevant investigations conducted on our patient

Name of investigations	Results	Reference values
Serum Calcium	5.6 mg/dL	8.1-10.6mg/dl
Serum Phosphate	2 mmol/L	Adult 0.8-1.6 mmol/L
Parathyroid Hormone (PTH)	70.1 pg/ml	15-65 pg/ml
Thyroid-Stimulating Hormone (TSH)	2.8 μIU/ml	0.4-4.2 μIU/ml
Follicle-Stimulating Hormone (FSH)	7.5 IU/L	4.5-21.5 IU/L
Luteinizing Hormone (LH)	10.1 IU/L	5-25 IU/L
Vitamin D	44 ng/mL	40 to 60 ng/mL
Serum Glutamate Pyruvate Transaminase (SGPT)	15 U/L	M< 41 U/L, F <31 U/L
Serum Albumin	4.3 g/dL	3.4 to 5.4 g/dL
Serum Creatinine	0.9mg/dL	M 0.6-1.4 mg/dL, F 0.6-1.2 mg/dL
Serum Sodium (Na)	134.1 mmol/L	135-145 mmol/L
Serum Potassium (K)	4.5 mmol/L	3.5-5.5 mmol/L
Serum Chloride (Cl)	103.2 mmol/L	98-108 mmol/L
Serum Magnesium	2 mg/dL	1.7 to 2.2 mg/dL
Random Blood Sugar (RBS)	70 mg/dl	< 140 mg/dl
Urine Calcium	190 mg/dl	50-250 mg/dl
Urine Phosphate	120 mg/dl	5-189 mg/dl

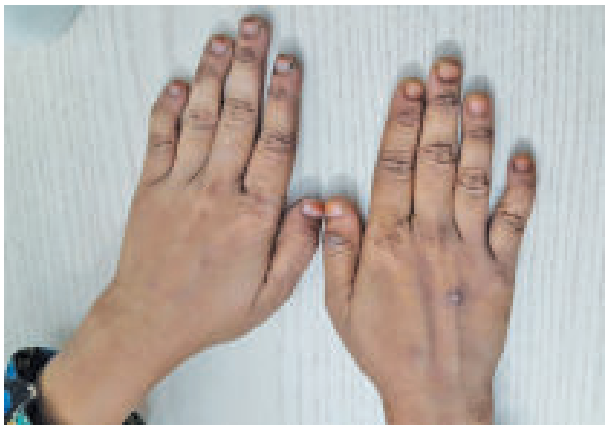

Figure 1: Feature of shortness of hands and brachydactyly

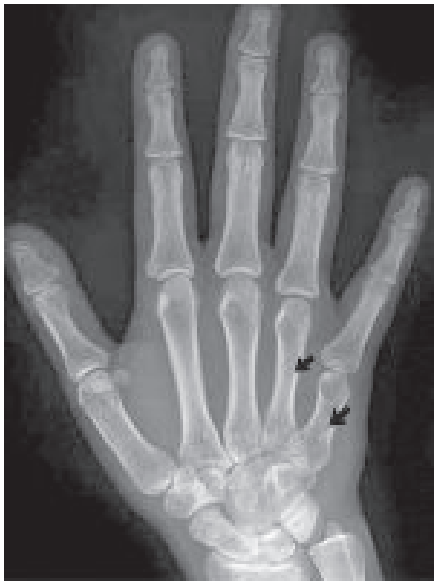

Figure 2: X-ray of hand showing shortness of fourth and fifth metacarpal bones

Discussion

Pseudohypoparathyroidism (PHP) is a heterogeneous group of disorders characterized by insensitivity to parathyroid hormone (PTH) in the targeted organs such as bone and kidney.[1] Pseudohypoparathyroidism (PHP) is a rare inherited disorder characterized by the body's reduced sensitivity or resistance to the effect of parathyroid hormone (PTH) at its target organs. Patients with the syndrome have hyperphosphatemia and hypocalcemia, which resemble hypoparathyroidism.[6] Pseudohypoparathyroidism is divided into subtypes, with PHP types 1a and 1b being the most common forms. These types are distinguished by the presence of physical features of Albright hereditary osteodystrophy (AHO) in PHP 1a, which were present in this patient. PHP1 and PHP2 have been traditionally distinguished based on the renal response to the exogenous PTH infusion, and PHP is diagnosed after other disorders have been ruled out. While cAMP is exclusively reduced in PHP1, urinary phosphate excretion is changed in both types. Without the AHO phenotype, the classification could be guided by resistance to TSH. The therapeutic behavior is unaffected by the PHP classification.[7] While parathyroid hormone-related peptide plays significant developmental roles, parathyroid hormone (PTH) is essential for controlling serum calcium and phosphate levels.[2]

Depending on the severity and chronicity of their condition, patients with PHP initially exhibit hypocalcemia signs and symptoms. Even when there is suggestive clinical evidence, the diagnosis of PHP is delayed since hypocalcemia is typically established as the primary diagnosis. After other illnesses have been ruled out, PHP is diagnosed. Differential diagnosis includes other conditions with a laboratory profile similar to PHP, such as renal disease, which can be easily ruled out by measuring creatinine, and osteomalacia.

Management involves calcium with dose modification until the calcium level falls within the normal range and vitamin D, ideally calcitriol supplementation to correct hypocalcemia and calcium level reaches normal limits under follow-up.[8] The treatment should be sufficiently strong to restore PTH levels and prevent the possible consequences of bone demineralization.[1] In this patient, ongoing monitoring of calcium and phosphate levels was recommended to guide therapy and prevent long-term complications such as nephrocalcinosis or soft tissue calcifications.

Target organ resistance to the hormone is the characteristic of PHP. This resistance is caused by a malfunction in the Gs-coupled receptor.[2] In another study, they found patients also have other hormone deficiencies using the same receptor and hypothyroidism and hypogonadism were present due to multiple hormone resistance. They treated patients with intravenous calcium replacement and then proceeded with oral calcium and vitamin D replacement.[3] Another study showed that a patient with a transient form of pseudohypoparathyroidism had significantly higher parathyroid hormone in the blood

but the patient had no hypocalcemia.[9] The objective is to keep thyroid function stable and ensure that calcium and phosphate levels in the blood remain within the normal range.

Findings that support PPH include hypocalcemia and hyperphosphatemia along with increased PTH and adequate renal function, even though genetic research is necessary for its rigorous classification. There are issues with the way that people with hypocalcemia are diagnosed, and it's possible that calcium problems are not often recognized as a differential diagnosis for common clinical manifestations including seizures and other neuromuscular symptoms. They also suggest that all patients with convulsive syndrome or basal ganglia calcifications must have a calcium issue ruled out.[10] A study shows due to G protein dysfunction, which results in resistance to TSH, LH, and FSH, PHP Type 1A is also linked to primary hypothyroidism and hypogonadism. Initially, patients have no goiter. Early in childhood, hypothyroidism may manifest before hypocalcemia does. Reproductive dysfunction is frequently observed in PHP Type 1A individuals.[4]

Genetic and cAMP testing could not be conducted due to limited local availability, emphasizing diagnostic challenges in low-resource settings. Despite the unavailability of confirmatory molecular diagnostics, the constellation of clinical features provided strong diagnostic confidence in our case of pseudohypoparathyroidism with AHO phenotype. This case demonstrates that a high index of clinical suspicion, combined with basic laboratory work and phenotypic features, can lead to the timely diagnosis and effective management of Pseudohypo-parathyroidism Type 1a, even in low-resource settings.

Conclusion

Pseudohypoparathyroidism and related disorders have a wide spectrum of clinical presentations which require a high degree of clinical suspicion to diagnose early. This case emphasizes the importance of considering pseudohypoparathyroidism in patients with hypocalcemia and elevated PTH levels. Early recognition and appropriate management are key to preventing complications associated with chronic hypocalcemia.

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Consent: Written informed consent was obtained from the patient to publish this case report in accordance with the journal's patient consent policy. A copy of the written consent is available for review by the editor-in-chief of this journal on request.

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