

Pseudohypoparathyroidism, Rare Cause of Hypocalcaemia!

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ABSTRACT

Pseudohypoparathyroidism is a rare disorder which is characterized by end organ parathormone resistance, which causes hypocalcaemia, hyperphosphataemia and high parathormone levels. We are reporting here case of a young male who had symptoms of chronic hypocalcaemia, with a positive Trousseau's and Chvostek's sign on examination, without any features of Albright's hereditary osteodystrophy. Lab investigations revealed low calcium, high phosphate and high PTH levels. The patient was diagnosed as having Pseudohypoparathyroidism and he was treated successfully with Calcium and Vitamin D supplements.

Key words: Tetany, Hypocalcaemia, Pseudohypoparathyroidism

CASE HISTORY

A 27-year-old male presented with intermittent spasms of both hands and feet, along with perioral and acral parasthaesias which he had since the past 10 years. Around 2 years back, in 2010, patient was diagnosed and unsatisfactorily treated for cervical dystonia. On examination, his vitals were found to be stable. Trousseau's and Chvostek sign was present. No distinctive skeletal or facial dysmorphism was present. He had a normal height of 168 cm.

On investigation, haemogram, urine examination, random blood glucose, renal function tests, serum sodium/potassium and liver function tests showed normal results. Serum calcium level was decreased, with a total calcium level of 5.1 mg/dl (8.4-10.2) and an ionised calcium level of 0.64 mmol/L (1.12-1.32). Serum Phosphorus was high 7.5 mg/dl (2.5-4.5). Serum Magnesium level was noted to be 1.8 mg/dl (1.6-2.3). Serum PTH (Intact) was high 186.7 pg/ml (12 to 65). Serum Vitamin D levels were normal (45.9). ECG, chest X-ray and a whole body skeletal survey showed normal results. Urinary c-AMP level and G_{α} subunit assay were not available.

He was managed initially with Calcium infusions and Vitamin D supplements. We discharged him on the high dose of oral Calcium (1gm elemental calcium/day) and Vitamin D (Calcitriol 60,000 IU/week). After 3 months, the tests were repeated and Total Calcium level was found to be 7.1 mg/dl (8.4-10.2) and Ionized Calcium level was found to be 0.82 mmol/L (1.12-1.32). The serum PTH level was persistently high and despite the patient being asymptomatic, clinical signs of tetany were still elicitable.

DISCUSSION

Pseudohypoparathyroidism is an uncommon disorder which is characterized by end organ Parathyroid Hormone (PTH) resistance [1]. In 1942, Fuller Albright first used the term, 'pseudohypoparathyroidism' to describe the PTH resistant hypocalcaemia and hyperphosphataemia, along with features of Albright hereditary osteodystrophy [2], which included developmental and skeletal defects. Since then, there has been an increased understanding of the differences at the levels of signal transduction and genetic expression [3]. A working classification of the various forms of PHP has been given in [Table/Fig-1].

Parasthaesias, numbness, tetany and convulsions are common presentations of severe hypocalcaemia. Classical clinical signs like Chvostek's and Trousseau's sign are useful pointers to the underlying electrolyte abnormalities. The threat of serious complications like

bronchospasms, laryngospasms, seizures and arrhythmias points out that a correct diagnosis and a prompt treatment are of utmost importance [5, 6].

Type	$\downarrow Ca^{+2}$ $\uparrow PO_4^-$	Urinary cAMP response to PTH	PTH	G_{α} Subunit Def	AHO	Other hormone resistance
PHP - Ia	YES	\downarrow	\uparrow	YES	YES	YES
PHP - Ib	YES	\downarrow	\uparrow	NO	NO	YES (few)
PHP - II	YES	NORMAL	\uparrow	NO	NO	No
PPHP	NO	NORMAL	NORMAL	YES	YES	No

[Table/Fig-1]: Types and features of pseudohypoparathyroidism.
PHP Pseudohypoparathyroidism, Ca^{+2} Calcium, PO_4^- Phosphate, PTH Parathormone
AHO Albrights Hereditary Osteodystrophy.

Treatment of PHP is similar to that of hypoparathyroidism, in the form of administration of oral Calcium and vitamin D or its analogues, except that Calcium and vitamin D doses are usually lower [1].

The presence of hypocalcaemia, hyperphosphataemia and high PTH levels without signs of Albright's hereditary osteodystrophy, points to a diagnosis of PHP of type Ib or type II. We treated this patient with oral Calcium and an active form of a Vitamin D analogue, and noted a partial clinical and laboratory response. He remains on close follow up, with a step up of doses and is on serial clinical and laboratory surveillance.

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