PSEUDOHYPOPARATHYROIDISM: FOUR CASES REPORTS.

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INTRODUCTION

Pseudohypoparathyroidism (PHP) is a rare disease characterized by a spectrum of disorders of mineral metabolism due parathyroid hormone (PTH) resistance. It can be classified in type 1a PHP (Albright Hereditary Osteodystrophy-OHA), PHP1b and PHP1c.

CASES REPORT

- ✓Four cases of PHP were identified, being 3 female.
- ✓The PHP diagnosis was performed at 6 years of age (in 3 cases) and at 7 years (in one case).
- ✓All had diagnosis of hypothyroidism, one had type 1 diabetes diagnosed at 12 years (6 years after PHP diagnosis) and another case had a mother with OHA phenotype but normal exams, characterizing pseudopseudohypoparathyroidism (PPHP).
- ✓Different clinical manifestations were observed such as craniofacial dysmorphisms (round face, flat nose ...), brachydactyly, short neck, mild mental retardation, subcutaneous calcifications, short stature and obesity.
- ✓Radiographic findings are suggestive of OHA in all cases: metacarpal shortening, epiphyseal anomalies, lytic lesions, subcutaneous calcifications or ossifications.
- ✓At diagnosis: all had elevated PTH. Other laboratory findings are presented in table 1.
- ✓All cases are being treated with 1,25 dihydroxy-vitamin D3 (calcitriol) replacement with or without oral calcium and await molecular analysis.

TABLE 1.

	PTH (pg/mL)	Total Calcium (mg/dL)	Phosphorus (mg/dL)
CASE 1	807.3	9.0	6.8
CASE 2	281.0	7.3	9.4
CASE 3	111.5	9.8	5.4
CASE 4	132.0	8.1	6.4
Reference values (RV)	15-65	8.2-10.3	4.0-7.0

DISCUSSION AND CONCLUSIONS

PHP is characterized by a defect in kidney response to PTH, with hyperphosphatemia and elevated PTH usually preceding hypocalcemia, being more commonly diagnosed in childhood. Genetically caused by mutations in the gene encoding G protein alpha subunit (GNAS), which signaling protein of PTH action, TSH, gonadotropins and ACTH, among others. All our cases were diagnosed in childhood and had clinical and radiological features of OHA.

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