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Pseudohypoparathyroidism; A rare cause of hypocalcemia

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A 14-year-old male patient presented to our endocrine outpatient clinic primarily with complaints of short stature. His height for age is below the –2SD. He had an episode of generalized tonic-clonic seizure 1 month back. Further evaluation revealed hypocalcemia. His biochemical investigations are mentioned below.

- Serum Corrected total calcium – 6.1mg/dl (8.5 – 10.3)
- Serum Phosphate – 5.6 mg/dl (2.5 – 4.5)
- Intact PTH – 250 pg/ml (20 – 80)

Figure 1 Short 4th and 5th metacarpals, metatarsals

What is the diagnosis?

1. Vitamin D deficienc
2. Primary hypoparathyroidism
3. **Pseudohypoparathyroidism Type 1a**
4. Pseudohypoparathyroidism Type 11
5. Pseudopseudohypoparathyroidism

Answer

Pseudohypoparathyroidism (PHP) refers to a group of heterogeneous disorders defined by target organ resistance (bone and kidney) to PTH (1). Typical Phenotype is called Albright hereditary osteodystrophy (AHO) which includes short stature, round face,

short 4th and 5th metacarpal and metatarsal bones, obesity (2). Biochemically PHP is characterized by hypocalcemia, hyperphosphatemia, and elevated PTH concentrations. But Different types of PHPshave different biochemical and phenotypical patterns which are mentioned in the table 1 below.

Table 1 different biochemical and phenotypical patterns

Type	AHO Phenotype	Biochemical phenotype
1. Pseudohypoparathyroidism Type 1a	Present	Present
2. Pseudohypoparathyroidism Type 1b	Absent	Present (Urine cyclic AMP low)
3. Pseudohypoparathyroidism Type 11	Absent	Present (Urine cyclic AMP high)
4. Pseudopseudohypoparathyroidism	Present	Absent

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