

BONES IN PSEUDOHYPOPARATHYROIDISM TYPE 1A

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Objectives:

Pseudohypoparathyroidism type 1a (PHP1a) is disorder characterised by resistance to the biological actions of circulating parathyroid hormone (PTH) along with typical features of Albright's hereditary osteodystrophy (AHO). Tissue-specific imprinting of GNAS gene results in complete resistance to PTH in renal proximal tubule, but skeletal responsiveness to PTH appears intact, since Gas is biallelically expressed in human bones.

The objective of this study was to determine BMD, and its relation to PTH levels in a group of young adult patients with PHP1a (age range 19-32yr).

Methods:

BMD measurements (Hologic QDR 4500) at the lumbar spine (LS), total hip (TH) and femoral neck (FN) were obtained in 5 patients with PHP1a (diagnosis was based on presence of AHO features combined with multihormone resistance). All patients were diagnosed with this condition earlier, and treatment was begun between ages 4 and 21 yr.

Results:

Characteristics of study patients: intact PTH levels ranged from 120.9 to 436pg/ml (reflecting inadequate dosing of calcium and calcitriol); all patients were taking levothyroxine and TSH levels were mildly elevated (range 5.98-12.10mIU/L); three subjects were GH deficient, and none received GH therapy.

The mean BMD Z-score for the LS was -1.4 (range -2.6 to -0.6), which is not significantly different from normal. Three subjects had Z-scores out of ± 1 , range expected for the normative population, and one of them met the criteria for low BMD. The mean BMD Z-score for the TH was -1.1 (range -1.6 to -0.8), and for the FN was -0.8 (range -1.9 to 0.2), both not significantly different from normal controls.

Conclusions:

Our results show that subjects with PHP1a, despite significant secondary hyperparathyroidism, had normal bone mass, with exception of one subject (20%) who had low LS BMD. These results are somewhat conflicting with previous study which demonstrated that bone mass is either normal or increased in PHP1a.

References:

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