Opis choroby *

Definicja

Pseudohypoparathyroidism type 2 (PHP2) is a type of pseudohypoparathyroidism (PHP; see this term) characterized by resistance to parathyroid hormone (PTH), which manifests with hypocalcemia, hyperphosphatemia and elevated PTH levels, absence of Albright's hereditary osteodystrophy (AHO; see this term), and normal expression of the Gs protein with a normal urinary cAMP response.

Dane

Klasyfikacja

Choroba

Kod ORPHA 94090

Kod OMIM 203330

Kod ICD10 E20.1

Kod ICD11 5A50.1

*Źródło

orphanet