

**DISEASE:**  
**Pseudohypoparathyroidism type 1B**

<b>NAME:</b>	Pseudohypoparathyroidism type 1B
<b>DESCRIPTION:</b>	Pseudohypoparathyroidism type 1B (PHP-1b) is a type of pseudohypoparathyroidism (PHP; see this term) characterized by localized resistance to parathyroid hormone (PTH) mainly in the renal tissues which manifests with hypocalcemia, hyperphosphatemia and elevated PTH levels. About 60-70% of patients also present with elevated TSH levels due to TSH resistance.
<b>ORPHACODE:</b>	94089
<b>XREF(S):</b>	<a href="#">Orphanet</a> <a href="#">MeSH</a> <a href="#">ICD-10</a> <a href="#">OMIM</a>
<b>ANALYTE(S):</b>	<a href="#">GNAS</a> <a href="#">STX16</a>
<b>CREATED:</b>	13 May 2019 - 01:02
<b>CHANGED:</b>	22 Jun 2023 - 16:14

## RELATED CONTENT

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### Related Genetic Tests

- Genetic disorders of Calcium and Phosphate metabolism (gene panel)
- Thyroid dysgenesis (38 genes)

### Related Laboratories

- Centrum Medische Genetica - UZ Brussel VUB
- Centrum Menselijke Erfelijkheid - KUL

### Related Analytes

- GNAS complex locus
- syntaxin 16

### Related Gene Panels

- Genetic disorders of Calcium and Phosphate metabolism (31 genes) - KUL
- Thyroid dysgenesis (38 genes) - VUB