

**DISEASE:**  
**Familial isolated hyperparathyroidism**

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| <b>NAME:</b>        | Familial isolated hyperparathyroidism                                                                                                                                                                                                                                                             |
| <b>DESCRIPTION:</b> | A rare, hereditary, familial primary hyperparathyroidism disease characterized by primary hyperparathyroidism due to single or multiple parathyroid tumors in at least two first-degree relatives in the absence of evidence of other endocrine disorders, tumors and/or systemic manifestations. |
| <b>ORPHACODE:</b>   | 99879                                                                                                                                                                                                                                                                                             |
| <b>SYNONYMS:</b>    | FIHPT                                                                                                                                                                                                                                                                                             |
| <b>XREF(S):</b>     | <a href="#">Orphanet</a><br><a href="#">OMIM</a><br><a href="#">MeSH</a><br><a href="#">OMIM</a><br><a href="#">OMIM</a><br><a href="#">OMIM</a><br><a href="#">ICD-10</a><br><a href="#">OMIM</a>                                                                                                |
| <b>ANALYTE(S):</b>  | <a href="#">GCM2</a><br><a href="#">CDC73</a><br><a href="#">MEN1</a>                                                                                                                                                                                                                             |
| <b>CREATED:</b>     | 13 May 2019 - 01:02                                                                                                                                                                                                                                                                               |

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| <b>CHANGED:</b> | 22 Jun 2023 - 16:14 |
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## RELATED CONTENT

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### Related Genetic Tests

- [Genetic disorders of Calcium and Phosphate metabolism \(gene panel\)](#)
- [Hyperparathyroidism \(gene panel\)](#)
- [Hyperparathyroidism \(gene panel\)](#)
- [Parathyroid tumor \(gene panel\)](#)

### Related Laboratories

- [Centre de Génétique Humaine - CHU Sart-Tilman](#)
- [Centrum Menselijke Erfelijkheid - KUL](#)

### Related Analytes

- [cell division cycle 73](#)
- [glial cells missing transcription factor 2](#)
- [menin 1](#)

### Related Gene Panels

- Genetic disorders of Calcium and Phosphate metabolism (31 genes) - KUL
- Hyperparathyroidism (3 genes) - KUL
- Hyperparathyroidism (3 genes) - KUL
- Parathyroid tumor (4 genes) - KUL

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