

Disease

# Familial isolated hypoparathyroidism 1<sup>1, 2, 3</sup>

Gene

## PTH

|                                                 |                                                                                                                                                                                                                                                     |
|-------------------------------------------------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>Inheritance</b>                              | AR and AD                                                                                                                                                                                                                                           |
| <b>Current Treatments</b>                       | calcium carbonate, calcitriol                                                                                                                                                                                                                       |
| <b>Evidence for Treatment</b>                   | expert opinion                                                                                                                                                                                                                                      |
| <b>Non-Molecular Testing Availability</b>       | yes                                                                                                                                                                                                                                                 |
| <b>Non-Molecular Test</b>                       | calcium, phosphate, parathyroid hormone <sup>2</sup>                                                                                                                                                                                                |
| <b>Experimental / Investigational Therapies</b> | <a href="https://clinicaltrials.gov/ct2/results?cond=Hypoparathyroidism&amp;term=&amp;cntry=&amp;state=&amp;city=&amp;dist=">https://clinicaltrials.gov/ct2/results?cond=Hypoparathyroidism&amp;term=&amp;cntry=&amp;state=&amp;city=&amp;dist=</a> |

References

- <sup>1</sup> <https://omim.org/entry/146200#13>
- <sup>2</sup> <https://pubmed.ncbi.nlm.nih.gov/28938448/>
- <sup>3</sup> <https://pubmed.ncbi.nlm.nih.gov/32207265/>