# DISEASE:
# Autosomal dominant hypocalcemia

| | |
|---|---|
| **NAME:** | Autosomal dominant hypocalcemia |
| **DESCRIPTION:** | A rare disorder of calcium homeostasis characterized by variable degrees of hypocalcemia with disproportionately low/normal levels of parathyroid hormone (PTH) and persistent normal or elevated renal calcium excretion. |
| **ORPHACODE:** | 428 |
| **SYNONYMS:** | AD hypocalcemia |
| **XREF(S):** | Orphanet<br>OMIM<br>OMIM<br>ICD-10 |
| **ANALYTE(S):** | CASR<br>GNA11 |
| **CREATED:** | 13 May 2019 - 01:02 |
| **CHANGED:** | 22 Jun 2023 - 16:14 |

# RELATED CONTENT

## Related Genetic Tests

- Endocrine Disorders - Hyper(Hypo)parathyroidism (gene panel - 24 genes)
- Genetic disorders of Calcium and Phosphate metabolism (gene panel)
- Hypocalciuric Hypercalcemia, Neonatal Severe Hyperparathyroidism, Hypocalcemia
- Hypocalciuric hypercalcemia, familial type I or Hypocalcemia or Hyperparathyroidism, familial isolated (CASR gene)
- Hypocalciuric hypercalcemia, familial type I or Hypocalcemia or Hypoparathyroidism, familial isolated (CASR gene)
- Parathyroid tumor (gene panel)
- Thyroid disgenesis (38 genes)
- Tubulopathy (gene panel)

## Related Laboratories

- Centre de Génétique Humaine - CHU Sart-Tilman
- Centre de Génétique Humaine - Erasme ULB
- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Medische Genetica - UZ Brussel VUB
- Centrum Menselijke Erfelijkheid - KUL

## Related Analytes

- calcium sensing receptor
- G protein subunit alpha 11

## Related Gene Panels

- Endocrine Disorders - Hyper(Hypo)parathyroidism (24 genes) - ULB
- Genetic disorders of Calcium and Phosphate metabolism (31 genes) - KUL
- Thyroid disgenesis (38 genes) - VUB
- Tubulopathy/Nephrolithiasis (106 genes) - IPG

---