

*Identify hypercalcemic individuals
who should not receive parathyroidectomy.*

Familial Hypocalciuric Hypercalcemia (CASR) Evaluation

What is Familial Hypocalciuric Hypercalcemia?

- Familial Hypocalciuric Hypercalcemia (FHH) is characterized by lifelong hypercalcemia that is caused by autosomal dominant loss-of-function mutations in the gene for the calcium-sensing receptor (CASR).¹
- FHH is generally asymptomatic and does not require treatment. Parathyroidectomy is not only unnecessary, but also ineffective.²
- Current diagnostic methods do not always allow reliable distinction between FHH and primary hyperparathyroidism, which is typically treated with parathyroidectomy.³

Why genetic testing?

Genetic testing for FHH-associated mutations in CASR

- Can confirm a suspected diagnosis of FHH and allow a definitive diagnosis of FHH in cases where biochemical tests are inconclusive.
- Can help to avoid an incorrect diagnosis of primary hyperparathyroidism that could lead to unnecessary and inappropriate parathyroidectomy in patients with FHH.
- Can identify FHH-associated mutations in relatives of FHH patients.

Indications for testing:

- Asymptomatic persistent hypercalcemia and normal or mildly elevated blood levels of parathyroid hormone
- Family history of FHH

*For complete ordering information,
please see the reverse side.*



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 **Correlagen**[®]
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www.athenadiagnostics.com

References: 1. Pollak MR et al (1993) *Cell* 75:1297-303. 2. Law WM Jr, Heath H 3rd (1985) *Ann Intern Med* 102:511-9. 3. Hendy GN et al (2000) *Human Mutation* 16:281-296.

For a brief review on FHH, please visit www.athenadiagnostics.com/DR

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Familial Hypocalciuric Hypercalcemia (CASR) Evaluation

- Typical Presentation:** Hypocalciuric hypercalcemia with inappropriately normal or mildly elevated blood levels of parathyroid hormone in individuals of any age
- Synonyms:** Familial benign hypercalcemia
- Indications for Testing:**
- Asymptomatic persistent hypercalcemia and normal or mildly elevated blood levels of parathyroid hormone
 - Family history of FHH

TEST DETAILS

- Test Code:** 829
- Test Turnaround:** 14-21 days

TECHNICAL INFORMATION

- Methodology:** Polymerase Chain Reaction (PCR), DNA sequencing of entire protein coding region of gene
- Patents:** United States Patent No.: 5,688,938

SHIPPING CONSIDERATIONS

- Specimen Type:** Whole blood
- Volume:** 10 mL (pediatric minimum: 2 mL)
- Collection Tube:** Yellow or lavender top
- Stability:** Hemolysis may compromise DNA recovery and integrity after 48 hrs
- Storage Conditions:** For short periods (until shipped) at 4°C
- Shipping Conditions:** Overnight at room temperature (specimen arrival must be less than 24 hrs after collection); ship Monday through Thursday only

Call Athena Diagnostics Customer Service Representatives to order the Familial Hypocalciuric Hypercalcemia (CASR) Evaluation (Test #829) at:

800-394-4493 x2



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