

GENETIC DISEASES EVALUATION CENTER

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Test Name & Code:

- **Test Name:** *CIAS1* Gene Mutations
- **Test Code:** 1849

Related Diseases & Genes:

- CINCA syndrome (OMIM #: 607115) || Gene: *NLRP3* (OMIM #: 606416)
- “Cold-induced autoinflammatory syndrome, familial” (OMIM #: 120100)
- Muckle-Wells syndrome (OMIM #: 191900)

Synonyms:

- *CIAS1* Gene (*CIAS1*)
- “Nacht Domain-, Leucine-Rich Repeat-, and PYD-Containing Protein 3 (NALP3)”
- Pyrin Domain-Containing APAF1-Like Protein 1 (PYPAF1)
- NALP3 Mutations
- PYPAF1 Mutations
- FCAS

Test Details:

- **Methodology:** DNA Sequencing
- **Performed:** Everyday
- **Reported:** 1-2 weeks

Specimen Details:

- **Specimen Collection:** Peripheral blood
- **Container:** Tube with EDTA
- **Specimen Volume/Amount:** > 5 ml
- **Storage/Transport Condition:** Refrigerated
- **Unacceptable Specimens/Conditions:** Serum; Frozen specimens; Hemolyzed specimens; Icterus; Lipemia

Testing Area(s):

- Multisystem Diseases

