
Reporting Title: GATA2 Gene, Full Gene Analysis**Performing Location:** Rochester**Ordering Guidance:**

For cases where the differential diagnosis remains broad, *GATA2* may be evaluated as part of a gene panel. See HLHGP / Primary Hemophagocytic Lymphohistiocytosis (HLH) Gene Panel, Varies; SCCNP / Severe Congenital and Cyclic Neutropenia Gene Panel, Varies; or EBLPD / Epstein Barr Virus (EBV) Susceptibility and Lymphoproliferative Disorders Gene Panel, Varies.

Targeted testing for familial variants (also called site-specific or known variants testing) is available for variants identified in the *GATA2* gene. See FMTT / Familial Variant, Targeted Testing, Varies. To obtain more information about testing option, call 800-533-1710.

Shipping Instructions:

Specimen preferred to arrive within 96 hours of collection.

Specimen Requirements:

Patient Preparation: A previous bone marrow transplant from an allogenic donor will interfere with testing. Call 800-533-1710 for instructions for testing patients who have received a bone marrow transplant.

Submit only 1 of the following specimens:

Specimen Type: Whole blood

Container/Tube:

Preferred: Lavender top (EDTA) or yellow top (ACD)

Acceptable: Any anticoagulant

Specimen Volume: 3 mL

Collection Instructions:

1. Invert several times to mix blood.
2. Send whole blood specimen in original tube. **Do not aliquot.**

Specimen Stability Information: Ambient (preferred) 4 days/Refrigerated

Specimen Type: Skin biopsy

Supplies: Fibroblast Biopsy Transport Media (T115)

Container/Tube: Sterile container with any standard cell culture media (eg, minimal essential media, RPMI 1640). The solution should be supplemented with 1% penicillin and streptomycin.

Specimen Volume: 4-mm punch

Specimen Stability Information: Refrigerated (preferred)/Ambient

Additional Information: A separate culture charge will be assessed under CULFB / Fibroblast Culture for Biochemical or Molecular Testing. An additional 3 to 4 weeks is required to culture fibroblasts before genetic testing can occur.

Specimen Type: Cultured fibroblasts

Container/Tube: T-25 flask

Specimen Volume: 2 Flasks

Collection Instructions: Submit confluent cultured fibroblast cells from a skin biopsy from another laboratory. Cultured

cells from a prenatal specimen will not be accepted.

Specimen Stability Information: Ambient (preferred)/Refrigerated (<24 hours)

Additional Information: A separate culture charge will be assessed under CULFB / Fibroblast Culture for Biochemical or Molecular Testing. An additional 3 to 4 weeks is required to culture fibroblasts before genetic testing can occur.

Forms:

1. **New York Clients-Informed consent is required.** Document on the request form or electronic order that a copy is on file. The following documents are available:
- [Informed Consent for Genetic Testing](#) (T576)
 - [Informed Consent for Genetic Testing \(Spanish\)](#) (T826)
2. [GATA2 Gene Sequencing Patient Information](#) (T811) is recommended.

Specimen Type	Temperature	Time	Special Container
Varies	Varies		

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
619803	Test Description	Alphanumeric		62364-5
619804	Specimen	Alphanumeric		31208-2
619805	Source	Alphanumeric		31208-2
619806	Result Summary	Alphanumeric		50397-9
619807	Result	Alphanumeric		82939-0
619808	Interpretation	Alphanumeric		69047-9
619809	Additional Results	Alphanumeric		82939-0
619810	Resources	Alphanumeric		99622-3
619811	Additional Information	Alphanumeric		48767-8
619812	Method	Alphanumeric		85069-3
619813	Genes Analyzed	Alphanumeric		82939-0
619814	Disclaimer	Alphanumeric		62364-5
619815	Released By	Alphanumeric		18771-6

LOINC® and CPT codes are provided by the performing laboratory.

Supplemental Report:

Supplemental

CPT Code Information:

- 81479
- 88233- Tissue culture, skin, solid tissue biopsy (if appropriate)
- 88240- Cryopreservation (if appropriate)

Test Definition: GATAS

GATA-Binding Protein 2, GATA2, Full Gene
Analysis, Next-Generation Sequencing, Varies

Reflex Tests:

Test Id	Reporting Name	CPT Units	CPT Code	Always Performed	Available Separately
CULFB	Fibroblast Culture for Genetic Test	1	88233	No	Yes

Result Codes for Reflex Tests:

Test ID	Result ID	Reporting Name	Type	Unit	LOINC®
CULFB	52327	Result Summary	Alphanumeric		50397-9
CULFB	52329	Interpretation	Alphanumeric		69965-2
CULFB	52328	Result	Alphanumeric		82939-0
CULFB	CG770	Reason for Referral	Alphanumeric		42349-1
CULFB	CG899	Specimen	Alphanumeric		31208-2
CULFB	52331	Source	Alphanumeric		31208-2
CULFB	52332	Method	Alphanumeric		85069-3
CULFB	54625	Additional Information	Alphanumeric		48767-8
CULFB	52333	Released By	Alphanumeric		18771-6

Reference Values:

An interpretive report will be provided