

Reporting Title: Dysregulation/Autoimmune GenePanel
Performing Location: Rochester

Ordering Guidance:
Targeted testing for familial variants (also called site-specific or known variants testing) is available for the genes on this panel. See FMTT / Familial Variant, Targeted Testing, Varies. To obtain more information about this 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. [Molecular Genetics: Congenital Inherited Diseases Patient Information](#) (T521)
3. [Inborn Errors of Immunity, Autoimmunity, and Autoinflammatory Disease Patient Information](#)

Specimen Type	Temperature	Time	Special Container
Varies	Varies		

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
619859	Test Description	Alphanumeric		62364-5
619860	Specimen	Alphanumeric		31208-2
619861	Source	Alphanumeric		31208-2
619862	Result Summary	Alphanumeric		50397-9
619863	Result	Alphanumeric		82939-0
619864	Interpretation	Alphanumeric		69047-9
619865	Additional Results	Alphanumeric		82939-0
619866	Resources	Alphanumeric		99622-3
619867	Additional Information	Alphanumeric		48767-8
619868	Method	Alphanumeric		85069-3
619869	Genes Analyzed	Alphanumeric		82939-0
619870	Disclaimer	Alphanumeric		62364-5
619871	Released By	Alphanumeric		18771-6

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

Supplemental Report:
Supplemental

CPT Code Information:

- 81443
- 88233-Tissue culture, skin, solid tissue biopsy (if appropriate)
- 88240-Cryopreservation (if appropriate)

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.