
Reporting Title: Tuberous Sclerosis Gene Panel**Performing Location:** Rochester**Ordering Guidance:**

Targeted testing (also called site-specific or known variant testing) is available for variants identified in this gene. See FMTT / Familial Variant, Targeted Testing, Varies.

Customization of this panel and single gene analysis for any gene present on this panel are available. See CGPH / Custom Gene Panel, Hereditary, Next-Generation Sequencing, Varies.

Additional Testing Requirements:

All prenatal specimens must be accompanied by a maternal blood specimen; order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on the maternal specimen as this must be a different order number than the prenatal specimen.

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. For instructions for testing patients who have received a bone marrow transplant, call 800-533-1710.

Submit only 1 of the following specimens:

Specimen Type: Whole blood

Container/Tube: 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)/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.

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.

Specimen Type: Blood spot

Supplies: Card-Blood Spot Collection (Filtration Paper) (T493)

Container/Tube:

Preferred: Collection card (Whatman Protein Saver 903 Paper)

Acceptable: PerkinElmer 226 (formerly Ahlstrom 226) filter paper or blood spot collection card (T493)

Specimen Volume: 5 Blood spots

Collection Instructions:

1. An alternative blood collection option for a patient older than 1 year is a fingerstick. For detailed instructions, see [How to Collect Dried Blood Spot Samples](#).
2. Let blood dry on the filter paper at ambient temperature in a horizontal position for a minimum of 3 hours.
3. Do not expose specimen to heat or direct sunlight.
4. Do not stack wet specimens.
5. Keep specimen dry.

Specimen Stability Information: Ambient (preferred)/Refrigerated

Additional Information:

1. Due to lower concentration of DNA yielded from blood spot, it is possible that additional specimen may be required to complete testing.
2. For collection instructions, see [Blood Spot Collection Instructions](#)
3. For collection instructions in Spanish, see [Blood Spot Collection Card-Spanish Instructions](#) (T777)
4. For collection instructions in Chinese, see [Blood Spot Collection Card-Chinese Instructions](#) (T800)

Specimen Type: Saliva

Patient Preparation: Patient should not eat, drink, smoke, or chew gum 30 minutes prior to collection.

Supplies: Saliva Swab Collection Kit (T786)

Specimen Volume: 1 Swab

Collection Instructions: Collect and send specimen per kit instructions.

Additional Information: Due to lower concentration of DNA yielded from saliva, it is possible that additional specimen may be required to complete testing.

Specimen Stability Information: Ambient 30 days

Prenatal Specimens

Due to its complexity, consultation with the laboratory is required for all prenatal testing; call 800-533-1710 to speak to a genetic counselor.

Specimen Type: Amniotic fluid

Container/Tube: Amniotic fluid container

Specimen Volume: 20 mL

Specimen Stability Information: Refrigerated (preferred)/Ambient

Additional information:

1. A separate culture charge will be assessed under CULAF / Culture for Genetic Testing, Amniotic Fluid.
2. All prenatal specimens must be accompanied by a maternal blood specimen; order MATCC / Maternal Cell

Contamination, Molecular Analysis, Varies on the maternal specimen.

Specimen Type: Chorionic villi
Container/Tube: 15-mL tube containing 15 mL of transport media
Specimen Volume: 20 mg
Specimen Stability Information: Refrigerated
Additional Information:
1. A separate culture charge will be assessed under CULFB / Fibroblast Culture for Biochemical or Molecular Testing.
2. **All prenatal specimens must be accompanied by a maternal blood specimen;** order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on the maternal specimen.

Acceptable:
Specimen Type: Confluent cultured cells
Container/Tube: T-25 flask
Specimen Volume: 2 flasks
Collection Instructions: Submit confluent cultured cells from another laboratory.
Specimen Stability Information: Ambient (preferred)/Refrigerated
Additional Information: **All prenatal specimens must be accompanied by a maternal blood specimen;** order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on the maternal specimen.

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: Neurology Patient Information](#)
3. If not ordering electronically, complete, print, and send one of the following with the specimen:
-[Neurology Specialty Testing Client Test Request](#) (T732)
-[Cardiovascular Test Request Form](#) (T724)

Specimen Type	Temperature	Time	Special Container
Varies	Varies		

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
616538	Test Description	Alphanumeric		62364-5
616539	Specimen	Alphanumeric		31208-2
616540	Source	Alphanumeric		31208-2
616541	Result Summary	Alphanumeric		50397-9
616542	Result	Alphanumeric		82939-0
616543	Interpretation	Alphanumeric		69047-9
616544	Resources	Alphanumeric		In Process
616545	Additional Information	Alphanumeric		48767-8

616546	Method	Alphanumeric		85069-3
616547	Genes Analyzed	Alphanumeric		48018-6
616548	Disclaimer	Alphanumeric		62364-5
616549	Released By	Alphanumeric		18771-6

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

Supplemental Report:

Supplemental

CPT Code Information:

- 81406
- 81407
- 81265-Maternal cell contamination (if appropriate)
- 88233-Tissue culture, skin, solid tissue biopsy (if appropriate)
- 88235-Amniotic Fluid culture (if appropriate)
- 88240-Cryopreservation (if appropriate)
- 81479 (if appropriate for government payers)

Reflex Tests:

Test Id	Reporting Name	CPT Units	CPT Code	Always Performed	Available Separately
MATCC	Maternal Cell Contamination, B	1	81265	No	Yes
CULAF	Amniotic Fluid Culture/Genetic Test	1	88235	No	Yes
CULFB	Fibroblast Culture for Genetic Test	1	88233	No	Yes
_STR1	Comp Analysis using STR (Bill only)	1	81265	No	No, (Bill only)
_STR2	Add'l comp analysis w/STR (Bill Only)	1	81266	No	No, (Bill only)

Result Codes for Reflex Tests:

Test ID	Result ID	Reporting Name	Type	Unit	LOINC®
CULAF	52304	Result Summary	Alphanumeric		50397-9
CULAF	52306	Interpretation	Alphanumeric		69965-2
CULAF	52305	Result	Alphanumeric		82939-0
CULAF	CG767	Reason for Referral	Alphanumeric		42349-1
CULAF	52307	Specimen	Alphanumeric		31208-2
CULAF	52308	Source	Alphanumeric		31208-2
CULAF	52309	Method	Alphanumeric		85069-3
CULAF	54641	Additional Information	Alphanumeric		48767-8
CULAF	52310	Released By	Alphanumeric		18771-6
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
MATCC	53285	Result Summary	Alphanumeric		50397-9
MATCC	53286	Result	Alphanumeric		40704-9
MATCC	53287	Interpretation	Alphanumeric		69047-9
MATCC	53288	Reason for referral	Alphanumeric		42349-1
MATCC	53289	Specimen	Alphanumeric		31208-2
MATCC	53290	Source	Alphanumeric		31208-2
MATCC	53291	Released By	Alphanumeric		18771-6
MATCC	55150	Method	Alphanumeric		85069-3

Reference Values:

An interpretive report will be provided.