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**Reporting Title: MSUD Gene Panel****Performing Location: Rochester****Ordering Guidance:**

The recommended first-tier screening tests for maple syrup urine disease (MSUD) are a combination of biochemical tests including quantitative plasma amino acids and urine organic acids. Order AAQP / Amino Acids, Quantitative, Plasma and OAU / Organic Acids Screen, Urine.

Customization of this panel and single gene analysis for any gene present on this panel is available. For more information see CGPH/ Custom Gene Panel, Hereditary, Generation Sequencing, Varies.

Targeted testing for familial variants (also called site-specific or known mutations 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. 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)

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 14 days

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 fibroblast

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.

Specimen Type: Blood spot

Supplies: Card-Blood Spot Collection (Filter 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

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.

Specimen Stability Information: Ambient 30 days

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

## **Specimen Minimum Volume:**

See Specimen Required

## **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 in:
  - Informed Consent for Genetic Testing (T576)
  - Informed Consent for Genetic Testing (Spanish) (T826)
2. Molecular Genetics: Biochemical Disorders Patient Information (T527)

| Specimen Type | Temperature | Time | Special Container |
|---------------|-------------|------|-------------------|
| Varies        | Varies      |      |                   |

**Ask at Order Entry (AOE) Questions:**

| Test ID | Question ID | Description         | Type       | Reportable |
|---------|-------------|---------------------|------------|------------|
| CULFB   | CG770       | Reason for Referral | Plain Text | No         |
| CULFB   | CG899       | Specimen            | Plain Text | No         |

**Result Codes:**

| Result ID | Reporting Name         | Type         | Unit | LOINC®  |
|-----------|------------------------|--------------|------|---------|
| 608776    | Test Description       | Alphanumeric |      | 62364-5 |
| 608777    | Specimen               | Alphanumeric |      | 31208-2 |
| 608778    | Source                 | Alphanumeric |      | 31208-2 |
| 608779    | Result Summary         | Alphanumeric |      | 50397-9 |
| 608780    | Result                 | Alphanumeric |      | 82939-0 |
| 608781    | Interpretation         | Alphanumeric |      | 69047-9 |
| 608782    | Resources              | Alphanumeric |      | 99622-3 |
| 608783    | Additional Information | Alphanumeric |      | 48767-8 |
| 608784    | Method                 | Alphanumeric |      | 85069-3 |
| 608785    | Genes Analyzed         | Alphanumeric |      | 48018-6 |
| 608786    | Disclaimer             | Alphanumeric |      | 62364-5 |
| 608787    | Released By            | Alphanumeric |      | 18771-6 |

LOINC and CPT codes are provided by the performing laboratory.

**Supplemental Report:**  
Supplemental**CPT Code Information:**

81405  
81406 x 3  
81479  
88233-Tissue culture, skin, solid tissue biopsy (if appropriate)  
88240-Cryopreservation (if appropriate)

81479 (if appropriate for government payers)

**Reflex Tests:**

| Test ID | Reporting Name                      | CPT Units | CPT Code | Always Performed | Orderable Separately |
|---------|-------------------------------------|-----------|----------|------------------|----------------------|
| CULFB   | Fibroblast Culture for Genetic Test |           |          | 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.