

NEW TEST ANNOUNCEMENT

NOTIFICATION DATE: October 3, 2013 **EFFECTIVE DATE:** October 08, 2013

MULTIPLE SULFATASE DEFICIENCY, KNOWN MUTATION Test ID: SUMFK

USEFUL FOR:

- Carrier testing of individuals with a family history of multiple sulfatase deficiency (MSD)
- Diagnostic confirmation of MSD when familial mutations have been previously identified

METHODOLOGY: Polymerase chain reaction (PCR) Amplification/DNA Sequencing

REFERENCE VALUES: An interpretive report will be provided.

SPECIMEN REQUIREMENTS:

- This test can only be performed if a mutation has previously been identified in a family member of this individual.
- Specimen must arrive within 96 hours of collection.

Submit only 1 of the following specimens:

Preferred:

Specimen Type: Whole blood

Container/Tube:

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

Acceptable: Any anticoagulant Specimen Volume: 3 mL Collection Instructions:

Invert several times to mix blood.
 Send specimen in original tube.

Specimen Stability Information: Ambient (preferred)/Refrigerated

Specimen Type: Cultured fibroblasts **Container/Tube:** T-75 or T-25 flask

Specimen Volume: 1 full T-75 or 2 full T-25 flasks

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

• Due to the complexity of prenatal testing, consultation with the laboratory is required for all prenatal testing.

Specimen Type: Amniotic fluid

Container/Tube: Amniotic fluid container

Specimen Volume: 20 mL

Specimen Stability Information: Refrigerated (preferred)/Ambient

Specimen Type: Chorionic villi

Container/Tube: 15-mL tube containing 15 mL of transport media

Specimen Volume: 20 mg

Specimen Stability Information: Refrigerated

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

Specimen Type: Skin biopsy

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. Tubes can be supplied upon request (Eagle's minimum essential medium with 1% penicillin and streptomycin [Supply T115]).

Specimen Volume: 4-mm punch

Specimen Stability Information: Refrigerated (preferred)/Ambient

Specimen Type: Blood spot

Container/Tube:

Preferred: Collection card (Whatman Protein Saver 903 Paper)

Acceptable: Ahlstrom 226 filter paper or Supplemental Newborn Screening Card

(Supply T493)

Specimen Volume: 2 to 5 blood spots on collection card

Collection Instructions:

- 1. An alternative blood collection option for a patient >1 year of age is finger stick.
- 2. Let blood dry on the filter paper at ambient temperature in a horizontal position for 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

NOTE:

- 1. Molecular Genetics-Biochemical Disorders Patient Information Sheet (Supply T527) in Special Instructions
- 2. New York Clients-Informed consent is required. Please document on the request form or electronic order that a copy is on file. An Informed Consent for Genetic Testing (Supply T576) is available in Special Instructions.

SPECIMEN STABILITY INFORMATION:

Specimen Type	Temperature	Time
Varies	Varies	

CAUTIONS:

- The identification of a disease-causing mutation in an affected family member is necessary before predictive testing for other family members can be offered. If a familial mutation has not been previously identified, order SUMFS/61816 SUMF1 Gene, Full Gene Analysis.
- Analysis is performed only for the provided familial mutations. This assay does not rule
 out the presence of other mutations within this gene or within other genes that may be
 associated with metabolic disease.

- Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Errors in our interpretation of results may occur if information given is inaccurate or incomplete.
- A previous bone marrow transplant from an allogenic donor will interfere with testing. Call Mayo Medical Laboratories at 800-533-1710 for instructions for testing patients who have received a bone marrow transplant.

CPT CODE:

81479-Unlisted molecular pathology procedure code

Additional tests:

Fibroblast Culture for Genetic Testing 88233-Tissue culture, skin, or solid tissue biopsy (if appropriate) 88240-Cryopreservation (if appropriate)

Amniotic Fluid Culture for Genetic Testing 88235-Tissue culture for amniotic fluid (if appropriate) 88240-Cryopreservation (if appropriate)

Maternal Cell Contamination, Molecular Analysis

81265-Comparative analysis using Short Tandem Repeat (STR) markers; patient and comparative specimen (eg, pre-transplant recipient and donor germline testing, post-transplant non-hematopoietic recipient germline [eg, buccal swab or other germline tissue sample] and donor testing, twin zygosity testing or maternal cell contamination of fetal cells (if appropriate)

DAY(S) SET UP: Tuesday; 10 a.m. ANALYTIC TIME: 10 days

QUESTIONS: Contact your Mayo Medical Laboratories' Regional Manager or Marvin H. Anderson, Jr., MML Laboratory Technologist Resource Coordinator Telephone: 800-533-1710