

NEW TEST ANNOUNCEMENT

NOTIFICATION DATE: September 19, 2012 **EFFECTIVE DATE:** September 26, 2012

FTCD Gene, Known Mutation

Test ID: GFDKM

USEFUL FOR:

- Diagnostic confirmation of glutamate formiminotransferase deficiency when familial mutations have been previously identified
- Carrier screening of individuals with a family history of glutamate formiminotransferase deficiency when mutation(s) in the *FTCD* gene have been identified in a family member
- Prenatal testing when 2 familial mutations have previously been identified in an affected family member

METHODOLOGY: Polymerase Chain Reaction (PCR) Followed by DNA Sequencing Analysis **REFERENCE VALUES**: An interpretive report will be provided.

SPECIMEN REQUIREMENTS: 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 flask or 2 full T-25 flasks

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

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

Acceptable:

Specimen Type: Blood spot

Container/Tube: Whatman Protein Saver 903 Paper

Specimen Volume: 5 blood spots

Collection Instructions:

- 1. Let blood dry on the filter paper at ambient temperature in a horizontal position for 3 hours.
- 2. Do not expose specimen to heat or direct sunlight.
- 3. Do not stack wet specimens.
- 4. Keep specimen dry.

Specimen Stability Information: Ambient (preferred)/Refrigerated

NOTE:

- Molecular Genetics-Biochemical Disorders Patient Information Sheet (Supply T527) in Special Instructions
- 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 GFDMS/89900 FTCD Gene, Full Gene Analysis.
- Analysis is performed for the known familial mutation(s) only. This assay does not rule out the presence of other mutations within this gene or within other genes that may be associated with mental retardation, high levels of serum folate, or high levels of urine formiminoglutamate.
- 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 1-800-533-1710 for instructions for testing patients who have received a bone marrow transplant.

FEE: Please contact your Regional Manager for your account's fee information.

CPT CODE: *FTCD* Gene, Known Mutation

83891-Isolation or extraction of highly purified nucleic acid

83898 x 2-Amplification, target, each nucleic acid sequence

83909 x 4-Separation and identification by high-resolution technique

83912-Interpretation and report

Fibroblast Culture for Genetic Testing

88233-Tissue culture, skin or solid tissue biopsy (if appropriate)

88240-Cryopreservation (if appropriate)

DAY(S) SET UP: Friday 10am **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