

NEW TEST ANNOUNCEMENT NOTIFICATION DATE: November 16, 2010 EFFECTIVE DATE: November 18, 2010

Ferrochelatase (FECH) Gene, Full Gene Analysis #60371

USEFUL FOR: Confirmation of a diagnosis of erythropoietic protoporphyria (EPP) following positive biochemical genetic test results obtained through #88886 "Porphyrins Evaluation, Erythrocytes"

Carrier testing for individuals with a family history of EPP in the absence of known mutations in the family

METHODOLOGY: Polymerase Chain Reaction (PCR) followed by DNA Sequence Analysis

REFERENCE VALUES: An interpretive report will be provided.

SPECIMEN REQUIREMENTS: Blood: 3 mL of whole blood lavender-top (EDTA) tube or yellow-top (ACD) tube

NOTE: Alternate: Blood Spots: 5 blood spots on "Whatman Protein Saver 903 Paper"

CAUTIONS:

- A small percentage of individuals who are carriers or have a diagnosis of erythropoietic protoporphyria (EPP) disease may have mutations that are not identified by this method (eg, large genomic deletions, promoter mutations). The absence of a mutation(s), therefore, does not eliminate the possibility of positive carrier status or the diagnosis of EPP disease. For carrier testing, it is important to first document the presence of a *FECH* gene mutation in an affected family member.
- This test does not exclude the presence of mutations within other genes, such as *ALAS2* (aminolevulinate, delta-, synthase 2), that are associated with EPP, X-linked or otherwise.
- In some cases, DNA alterations of undetermined significance may be identified.
- Rare polymorphisms exist that could lead to false-negative or false-positive results. If results obtained do not match the clinical findings, additional testing should be considered.
- A previous bone marrow transplant from an allogenic donor will interfere with testing. Call Mayo Medical Laboratories for instructions for testing patients who have received a bone marrow transplant.
- 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.

LIST FEE: \$1092.00

CPT CODE: 83891-Isolation or extraction of highly purified nucleic acid 83898 x 11-Amplification, target, each nucleic acid sequence 83909 x 22-Separation and identification by high-resolution technique 83912-Interpretation and report

ANALYTIC TIME: 5 days

DAY(S) SET-UP:

Wednesdays, 10 AM

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