

NEW TEST ANNOUNCEMENT

NOTIFICATION DATE: August 30, 2012 **EFFECTIVE DATE:** August 30, 2012

Gelsolin (GSN) Gene, Full Gene Analysis Test ID: GSNMS

USEFUL FOR: Diagnostic confirmation of amyloidosis V

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

REFERENCE VALUES: An interpretive report will be provided.

SPECIMEN REQUIREMENTS: Specimen must arrive within 96 hours of draw.

Container/Tube:

Preferred: 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 specimen in original tube.

Note:

- Molecular Genetics-Congenital Inherited Diseases Patient Information Sheet (Supply T521)
- 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	Ambient (preferred)	
	Frozen	
	Refrigerated	

CAUTIONS:

- A small percentage of individuals who have a diagnosis of amyloidosis V may have a mutation that is not identified by this method (eg, large genomic deletions, promoter mutations). The absence of a mutation, therefore, does not eliminate the possibility of positive carrier status or the diagnosis of amyloidosis V. For carrier testing, it is important to first document the presence of a gelsolin (*GSN*) gene mutation in an affected family member.
- 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.
- Mutations in other genes, such as those encoding transthyretin, fibrinogen alpha chain, apolipoprotein AII, lysozyme and others have been shown to cause other forms of familial amyloidosis. Abnormalities in these genes are not detected by this assay.

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

CPT CODE:

83890-Isolation or extraction of highly purified nucleic acid 83898 x 17-Amplification, target, each nucleic acid sequence 83909 x 34-Separation and identification by high-resolution technique 83912-Interpretation and report

DAY(S) SET UP: Friday, 10 am

ANALYTIC TIME: 10 days