

NEW TEST ANNOUNCEMENT

NOTIFICATION DATE: October 5, 2012 **EFFECTIVE DATE:** October 11, 2012

CDKN1C Gene, Full Gene Analysis Test ID: CDKMS

USEFUL FOR:

- Confirming a clinical diagnosis of Beckwith-Wiedemann syndrome
- Confirming a clinical diagnosis of IMAGe syndrome

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 collection.

Submit only 1 of the following specimens:

Specimen Type: Blood

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.

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 (egminimal 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

NOTE:

- Molecular Genetics-Congenital Inherited Diseases Patient Information Sheet (Supply T521) 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:

- A small percentage of individuals who are carriers or have a diagnosis of Beckwith-Wiedemann syndrome or IMAGe syndrome caused by *CDKN1C* 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 Beckwith-Wiedemann syndrome or IMAGe syndrome. For carrier testing, it is important to first document the presence of a *CDKN1C* 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 at 1-800-533-1710 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.

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

CPT CODE:

CDKN1C Gene, Full Gene Analysis
83891- Isolation or extraction of highly purified nucleic acid
83898 x 6- Amplification, target, each nucleic acid sequence
83909 x 12- 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, 10 am

ANALYTIC TIME: 10 days

NOTE: The following referral test code(s) will become obsolete.

Test Name	Test ID	Referral Lab Code	Referral Lab
Beckwith Wiedemann	ZW169	MML891	Univ of Pennsylvania
Syndrome (BWS),			School of Medicine
CDKN1C Sequencing			
Analysis			

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