MAYO
Mayo Medical Laboratories
**1-800-533-1710**

**NEW TEST ANNOUNCEMENT**
**NOTIFICATION DATE:** December 24, 2013
**EFFECTIVE DATE:** January 3, 2014

# CHEK2 Gene, Full Gene Analysis
Test ID: CHEKS

**USEFUL FOR**:
- Evaluation for hereditary susceptibility to breast cancer or Li-Fraumeni-like syndrome
- Identification of a familial CHEK2 mutation to allow for predictive testing in family members

**METHODOLOGY**: Polymerase Chain Reaction (PCR) Amplification/DNA Sequencing Array, comparative genomic hybridization (aCGH) is used to test for the presence of large deletions and duplications

**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-Colon Cancer 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 | Ambient (preferred) | |
| | Frozen | |
| | Refrigerated | |

**CAUTIONS:**
- Some individuals who have an inherited susceptibility to breast cancer and other cancers may have a mutation in CHEK2 that is not identified by this method (eg, deep intronic mutations, promoter mutations). The absence of a mutation, therefore, does not eliminate the possibility of an inherited susceptibility to breast or other cancers. For predictive testing of asymptomatic individuals, it is important to first document the presence of a CHEK2 gene mutation in an affected family member.

- In some cases, DNA alterations of undetermined significance may be identified.
- We strongly recommend that asymptomatic patients undergoing predictive testing receive genetic counseling both prior to testing and after results are available.
- Predictive testing of an asymptomatic child is not recommended.
- 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 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.

**CPT CODE**:
81479-Unlisted molecular pathology code

Hereditary Colon Cancer CGH Array
81228-Cytogenomic constitutional (genome-wide) microarray analysis; interrogation of genomic regions for copy number variants (eg, Bacterial Artificial Chromosome [BAC] or oligo-based comparative genomic hybridization [CGH] microarray analysis)

**DAY(S) SET UP**: Wednesday, 10 a.m. **ANALYTIC TIME**: 14 days

NOTE: The following referral test code(s) will become obsolete effective February 3, 2014.

| Test Name | Test ID | Referral Lab Code | Referral Lab |
|---|---|---|---|
| CHEK2 (Checkpoint Kinase 2) Gene Mutation Analysis - Full gene sequencing | ZW171 | CHEK2-SEQ | City of Hope National Medical Center |
| CHEK2 gene, MLPA analysis - Deletion | ZW171 | CHEK2-DEL | City of Hope National Medical Center |

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