



Single-Gene Large Deletion and Duplication Analysis
Test ID: SDEL

USEFUL FOR: Diagnostic or predictive testing for hereditary colorectal cancer when a large deletion or duplication in 1 of the following genes has been identified in a family member; AXIN2, BMPR1A, CDH1, CHEK2, MLH3, TP53, PMS2, PTEN, SCG5-GREM1, SMAD4, and STK11

METHODOLOGY: Gene Dosage Analysis by Array Comparative Genomic Hybridization (aCGH)

REFERENCE VALUES: An interpretive report will be provided.

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

Specimen Type: Whole 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.
3. To ensure minimum volume and concentration of DNA are met, **3 mL of blood must be submitted.** Testing may be canceled if DNA requirements are inadequate.

NOTE:

- 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.
- Molecular Genetics-Colon Cancer Patient Information Sheet (Supply T521) in Special Instructions

SPECIMEN STABILITY INFORMATION:

Specimen Type	Temperature	Time
Varies	Ambient (preferred)	
	Frozen	
	Refrigerated	

CAUTIONS:**Clinical Correlations**

- 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, contact Mayo Medical Laboratories at 800-533-1710 to discuss testing options.
- Test results should be interpreted in context of clinical findings, family history, and other laboratory data. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.

Technical Limitations

- In some cases, DNA variants 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 800-533-1710 for instructions for testing patients who have received a bone marrow transplant.

Evaluation Tools

- Multiple in-silico evaluation tools were used to assist in the interpretation of these results. These tools are updated regularly; therefore, changes to these algorithms may result in different predictions for a given alteration. Additionally, the predictability of these tools for the determination of pathogenicity is currently unvalidated.

Reclassification of Variants-Policy

- All detected alterations are evaluated according to American College of Medical Genetics and Genomics recommendations. Variants are classified based on known, predicted, or possible pathogenicity and reported with interpretive comments detailing their potential or known significance. At this time, it is not standard practice for the laboratory to systematically review likely deleterious alterations or variants of uncertain significance that are detected and reported. The laboratory encourages health care providers to contact the laboratory at any time to learn how the status of a particular variant may have changed over time.

CPT CODE:

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: Varies, weekly to every other week based on sample volume, will be run twice a month at minimum.

ANALYTIC TIME: 3 weeks

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