MAYO
Mayo Medical Laboratories
1-800-533-1710

NEW TEST ANNOUNCEMENT
NOTIFICATION DATE: November 26, 2013
EFFECTIVE DATE: December 3, 2013

NOONAN SPECTRUM SEQUENCE PANEL 1 FOR *KRAS*, *PTPN11*, *RAF1*, *SOS1*, BLOOD
Test ID: NS1

USEFUL FOR: Aiding in the diagnosis of Noonan syndrome and related phenotypes such as LEOPARD syndrome

GENETICS INFORMATION: This test involves simultaneous sequence analysis of the four genes most commonly involved in Noonan syndrome. This test is more cost effective than ordering each of the four genes individually. Please call 1-800-533-1710 and ask to speak to the genetic counselor if you have questions about testing strategy for Noonan syndrome.

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

REFERENCE VALUES: An interpretive report will be provided.

SPECIMEN REQUIREMENTS:
Multiple gene sequencing tests can be performed on a single specimen after a single extraction. See Multiple Cardiovascular-Related Gene Sequencing Tests in Special Instructions for a list of tests that can be ordered together.

Container/Tube: Lavender top (EDTA)
Specimen Volume: 3 mL
Collection Instructions: Send specimen in original tube.
Additional Information:

1. Include physician's name and phone number with the specimen.
2. Transfusions will interfere with testing for up to 4 to 6 weeks. DNA obtained from white cells may not provide useful information for patients who received a recent transfusion of blood that was not leukocyte reduced. Wait 4 to 6 weeks until transfused cells have left the patient's circulation before drawing the patient's blood specimen for genotype testing.

Forms:

1. Required for all orders: Noonan Spectrum Gene Testing Patient Information Sheet (Supply T689) in Special Instructions
2. 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.

Minimum Volume: 0.5 mL

SPECIMEN STABILITY INFORMATION:

Specimen Type	Temperature	Time
Whole Blood EDTA	Ambient (preferred)	
	Refrigerated	

CAUTIONS:

- Absence of a mutation does not preclude the diagnosis of Noonan syndrome or another related disorder. Additional genes not tested by this assay known to be involved in Noonan syndrome and related phenotypes include NRAS, HRAS, BRAF, SHOC2, MAP2K1, MAP2K2, and CBL.
- This method will not detect mutations that occur in the introns (except in the splicing regions) and regulatory regions of the gene and large rearrangement-type mutations.
- Sometimes a genetic alteration of unknown significance may be identified. In this case, testing of appropriate family members may be useful to determine pathogenicity of the alteration.

CPT CODE:

81405 - *KRAS* (v-Ki-ras2 Kirsten rat sarcoma viral oncogene homolog) (e.g., Noonan syndrome), full gene sequence

81406 - *PTPN11* (protein tyrosine phosphatase, non-receptor type 11) (e.g., Noonan syndrome, LEOPARD syndrome), full gene sequence

81404 - *RAF1* (v-raf-1 murine leukemia viral oncogene homolog 1) (e.g., LEOPARD syndrome), full gene sequence

81406 - *SOS1* (son of sevenless homolog 1) (e.g., Noonan syndrome, gingival fibromatosis), full gene sequence

DAY(S) SET UP: varies

ANALYTIC TIME: 7 days

QUESTIONS: Contact your Mayo Medical Laboratories' Regional Manager or
Shirley Pokorski, MML Laboratory Technologist Resource Coordinator
Telephone: 800-533-1710