

NEW TEST ANNOUNCEMENT

NOTIFICATION DATE: November 26, 2013 **EFFECTIVE DATE:** December 3, 2013

SOS1, FULL GENE SEQUENCE, BLOOD Test ID: SOS1

USEFUL FOR: Aiding in the diagnosis of *SOS1*-associated Noonan syndrome and hereditary gingival fibromatosis

GENETICS INFORMATION: This test aids in the diagnosis of *SOS1*-associated Noonan syndrome and related clinical disorders.

PROFILE INFORMATION:

Test ID	Reporting Name	Available Separately	Always Performed
SOS1F	SOS1, Full Gene Sequence	No	Yes

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:

- Related genetic tests available include testing for a specific familial mutation (SOSK / SOSI Gene, Known Mutation, Blood), which should be used when testing individuals who are at risk for a SOSI mutation that has been previously identified in the family.
- Absence of a mutation does not preclude the diagnosis of Noonan syndrome or another *SOS1*-related disorder unless a specific mutation has already been identified in an affected family member.
- 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:

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

NOTE: The following referral test code(s) will become obsolete effective December 3, 2013.

Test Name	Test ID	Referral Lab Code	Referral Lab
Noonan Syndrome via SOS1 Gene	ZW194	376	Prevention Genetics Lab
Sequencing			

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