

Patient Name TESTINGRNV,SOSK POS	Patient ID SA00064127	Age 12	Gender M	Order # SA00064127
Ordering Phys CLIENT,CLIENT			DOB 11/12/2000	
Client Order # SA00064127	Account Information			Report Notes
Collected 11/05/2013 12:00	C7028846-DLMP Rochester SDSC 2 - Client Support Rochester, MN 55901			
Printed 11/26/2013 13:02				

Test	Flag	Results	Unit	Reference Value	Perform Site*
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SOS1 Gene, Known Mutation, B
RECEIVED: 11/06/2013 09:18 **REPORTED:** 11/07/2013 08:50

SOS1 Gene, Known Mutation

SOS1 Known Mutation Result

MCR

The following heterozygous SOS1 familial variant was detected in this individual: Exon 10, nucleotide c.1300G>C, amino acid p.Gly434Arg (p.G434R).

SOS1 Known Mut Interpretation

MCR

The SOS1 p.Gly434Arg (p.G434R) variant was previously identified in a family member with features of Noonan syndrome (NS). The presence of this variant, therefore, suggests that this individual is at risk for development and/or exacerbation of features of NS. Appropriate surveillance procedures and/or management strategies should be considered.

Since a variant has been identified in the SOS1 gene in this individual, genetic testing for this specific variant in other family members is recommended. Please contact the laboratory at 1-800-533-1710 or the on-line test catalog at www.mayomedicallaboratories.com for information about how to order the test for SOSK (SOS1 Gene, Known Mutation, B). Please refer to family number 12345 when ordering testing on family members of this individual. Fluorescent DNA sequence analysis was used to test for the presence of a specific sequence change in the SOS1 gene (GenBank accession number NM_005633.3), which was previously identified in an affected family member of this individual.

A genetic consultation may be of benefit.

CAUTIONS:

Rare polymorphisms exist that could lead to false negative or positive results. If results obtained do not match the clinical findings, additional testing should be considered.

Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.

Bone marrow transplants from allogenic donors will interfere with testing. Call Mayo Medical Laboratories for

Performing Site Legend on Last Page of Report

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* Report times for Mayo performed tests are CST/CDT

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instructions for testing patients who have received a bone marrow transplant. Laboratory developed test. Reviewed By: Linnea M. Baudhuin, Ph.D. See Below MCR Result:					

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director:
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