

Patient ID SA00064127	Patient Name TESTINGRNV, SOSK POS	Birth Date 2000-11-12	Gender M	Age 12
Order Number SA00064127	Client Order Number SA00064127	Ordering Physician Client, Client	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 05 Nov 2013 12:00		

SOS1 Gene, Known Mutation, B

SOS1 Gene, Known Mutation

SOS1 Known Mutation Result

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

MCR

(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.

If the patient has had an allogeneic blood or marrow transplant or a recent (i.e. less than 6 weeks from time of sample collection) heterologous blood transfusion these results may be inaccurate due to the presence of donor DNA. Laboratory developed test.

Reviewed By

MCR

Linnea M. Baudhuin, Ph.D.

Received: 06 Nov 2013 09:18

Reported: 07 Nov 2013 08:50

SOS1 Known Mut Interpretation

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.

MCR

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.

ADDITIONAL INFORMATION

Fluorescent DNA sequence analysis was used to test for the presence of a specific sequence change in the SOS1 gene

Performing Site Legend

Code	Laboratory	Address
MCR	Mayo Clinic Dept. of Lab Med and Pathology	200 First Street SW, Rochester, MN 55905