



Patient ID <b>SA00064126</b>	Patient Name <b>TESTINGRNV, SOSK NEG</b>	Birth Date <b>2000-03-11</b>	Gender <b>U</b>	Age <b>13</b>
Order Number <b>SA00064126</b>	Client Order Number <b>SA00064126</b>	Ordering Physician <b>Client, Client</b>	Report Notes	
Account Information <b>C7028846 DLMP Rochester</b>		Collected <b>05 Nov 2013 12:00</b>		

## SOS1 Gene, Known Mutation, B

### SOS1 Gene, Known Mutation

#### SOS1 Known Mutation Result

MCR

The SOS1 Exon 10, nucleotide c.1300G>C, amino acid p.Gly434Arg (p.G434R) familial variant was not detected in this individual.

#### SOS1 Known Mut Interpretation

MCR

The SOS1 p.Gly434Arg (p.G434R) variant was previously identified in a family member with features of Noonan syndrome. Since this variant was not detected in this individual, this suggests that this patient is at no greater risk than someone in the general population for having Noonan syndrome.

#### ADDITIONAL INFORMATION

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.

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 08:49

#### Performing Site Legend

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