

Laboratory Service Report

1-800-533-1710

Patient Name TESTINGRNV,NS1	Patient ID SA00064199	Age 33	Gender F	Order # SA00064199
Ordering Phys CLIENT,CLIENT				DOB 01/21/1980
Client Order # SA00064199	Account Information C7028846-DLMP Rochester SDSC 2 - Client Support		Report Notes	
Collected 11/06/2013 09:59				
Printed 11/26/2013 12:46	Rochester, MN 55901			

Test	Flag Results	Unit	Reference Value	Perform Site*
Noonan Spectrum Sequence Panel 1, B RECEIVED: 11/07/2013 12:45 REPORTED	D: 11/07/2013 15:59			MCD
A heterozygous pathogenic va Exon 14, nucleotide c.145663 (p.D486N). A pathogenic var	ariant was detected in R >A, amino acid p.Asp486A Lant was not detected in	AF1: sn the		MCK
other genes analyzed in this	s panel.			
Noonan Spectrum Interpretation	1 (175)			MCR
The presence of a Noonan syn	ndrome (NS) - associated			
pathogenic variant in this p	patient places this indi	vidual		
at risk for developing and/	or exacerbation of sympt	oms		
associated with this condit:	Lon. Appropriate surveil	lance		
procedures and/or management	t strategies should be			
considered.				
Since a nathogenic variant l	as been identified in t	he PAF1		
gene in this individual de	as been identified in t	necific		
variant in other family mem	pers is recommended Ple	250 250		
contact the laboratory at 1-	-800-533-1710 or the on-	line		
test catalog at www.mayomed	icallaboratories.com for	11110		
information about how to or	der the test RAF1K (RAF1	Gene.		
Known Mutation, B). Please	refer to family number X	YZ when		
ordering testing on family r	nembers of this individu	al.		
Fluorescent DNA sequence and	alvsis was used to test	for the		
presence of mutations in all	L coding exons (1-15) an	d		
corresponding exon-intron be	oundaries of the PTPN11	gene		
(GenBank accession number NI	4 002834.3), all coding	exons		
(2-17) and corresponding exe	on-intron boundaries of	the RAF1		
gene (GenBank accession num)	per NM 002880.3), all co	ding		
exons (1-23) and correspond:	ing exon-intron boundari	es of		
the SOS1 gene (GenBank acces	ssion number NM 005633.3), and		
all coding exons (2-6 (Varia	ant A)) and correspondin	g		
exon-intron boundaries of th	ne KRAS gene (GenBank ac	cession		
number NM 033360.2).				
Laboratory developed test.				
Reviewed by	See Below			MCR
Result:Linnea M. Baudhuin, 1	Ph.D.			

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director: Franklin R. Cockerill, III, M.D.

Patient Name TESTINGRNV,NS1	Collection Date and Time 11/06/2013 09:59	Report Status Final		
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* Demonstration of four Marian and family of the star and OOT/ODT				

Report times for Mayo performed tests are CST/CDT