

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			Report Notes
Collected 11/06/2013 09:59	C7028846-DLMP Rochester SDSC 2 - Client Support Rochester, MN 55901			
Printed 11/26/2013 12:46				

Test	Flag	Results	Unit	Reference Value	Perform Site*
Noonan Spectrum Sequence Panel 1, B					
RECEIVED: 11/07/2013 12:45 REPORTED: 11/07/2013 15:59					
Noonan Spectrum Panel Result					
A heterozygous pathogenic variant was detected in RAF1: Exon 14, nucleotide c.1456G>A, amino acid p.Asp486Asn (p.D486N). A pathogenic variant was not detected in the other genes analyzed in this panel.					
Noonan Spectrum Interpretation					
The presence of a Noonan syndrome (NS) - associated pathogenic variant in this patient places this individual at risk for developing and/or exacerbation of symptoms associated with this condition. Appropriate surveillance procedures and/or management strategies should be considered.					
Since a pathogenic variant has been identified in the RAF1 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 RAF1K (RAF1 Gene, Known Mutation, B). Please refer to family number XYZ when ordering testing on family members of this individual. Fluorescent DNA sequence analysis was used to test for the presence of mutations in all coding exons (1-15) and corresponding exon-intron boundaries of the PTPN11 gene (GenBank accession number NM_002834.3), all coding exons (2-17) and corresponding exon-intron boundaries of the RAF1 gene (GenBank accession number NM_002880.3), all coding exons (1-23) and corresponding exon-intron boundaries of the SOS1 gene (GenBank accession number NM_005633.3), and all coding exons (2-6 (Variant A)) and corresponding exon-intron boundaries of the KRAS gene (GenBank accession number NM_033360.2). Laboratory developed test.					
Reviewed by		See Below			MCR
Result:Linnea M. Baudhuin, Ph.D.					

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director:
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Patient Name TESTINGRNV,NS1	Collection Date and Time 11/06/2013 09:59	Report Status Final
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* Report times for Mayo performed tests are CST/CDT