MAYO CLINIC
Mayo Medical Laboratories

1-800-533-1710

Patient ID	Patient Name		Birth Date	Gender	Age
SA00043163	SAMPLEREPORT, PT11		1966-06-10	F	45
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00043163	SA00043163	Client, Client			
Account Information		**Collected**			
C7028846 DLMP Rochester		07 Feb 2012 13:00			

DNA Extraction, NPL

Specimen MCR

Blood

Specimen ID MCR

1037724

Order Date MCR

09 Feb 2012 07:23

Comment MCR

Genomic DNA has been extracted.

Reviewed By MCR

Melody Elizabeth Kimball

Release Date MCR

09 Feb 2012 07:24

Received: 08 Feb 2012 16:02 **Reported:** 09 Feb 2012 07:25

Performing Site Legend

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

Patient ID	Patient Name		Birth Date	Gender	Age
SA00043163	SAMPLEREPORT, PT11		1966-06-10	F	45
Order Number	Client Order Number	Ordering Physician	Report Notes		
SA00043163	SA00043163	Client, Client			
Account Information		Collected			
C7028846 DLMP Rochester		07 Feb 2012 13:00			

PTPN11, Full Gene Sequence, B

PTPN11, Full Gene Sequence

PTPN11 Full Gene Result

MCR

One copy of the following mutation was detected in PTPN11:
Exon 3, nucleotide c.236A>G, amino acid p.Gln79Arg (p.G79R).

PTPN11 Full Gene Interpretation

MCR

This PTPN11 mutation is consistent with the phenotypic features observed in this patient. Appropriate surveillance and management strategies should be considered.

Since a mutation has been identified in the PTPN11 gene in this individual, genetic testing for this specific mutation 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 PTPN11 Gene, Known Mutation, B (89464). Please refer to family number 123456 when ordering testing on family members of this individual.

ADDITIONAL INFORMATION

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).

A genetic consultation may be of benefit.

A list of common polymorphisms identified for this patient is available from the lab upon request.

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

Jamie Bruflat

PTPN11, Full Gene Sequencing

MCR

Performed

Received: 08 Feb 2012 16:02 **Reported:** 08 Feb 2012 16:07

Performing Site Legend

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