



Patient ID SA00043484	Patient Name SAMPLEREPORT, PT1K	Birth Date 1966-06-10	Gender F	Age 45
Order Number SA00043484	Client Order Number SA00043484	Ordering Physician Client, Client	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 09 Feb 2012 13:00		

DNA Extraction, NPL

Specimen

Blood

MCR

Comment

Genomic DNA has been extracted.

MCR

Specimen ID

1037736

MCR

Reviewed By

Melody Elizabeth Kimball

MCR

Order Date

10 Feb 2012 10:28

MCR

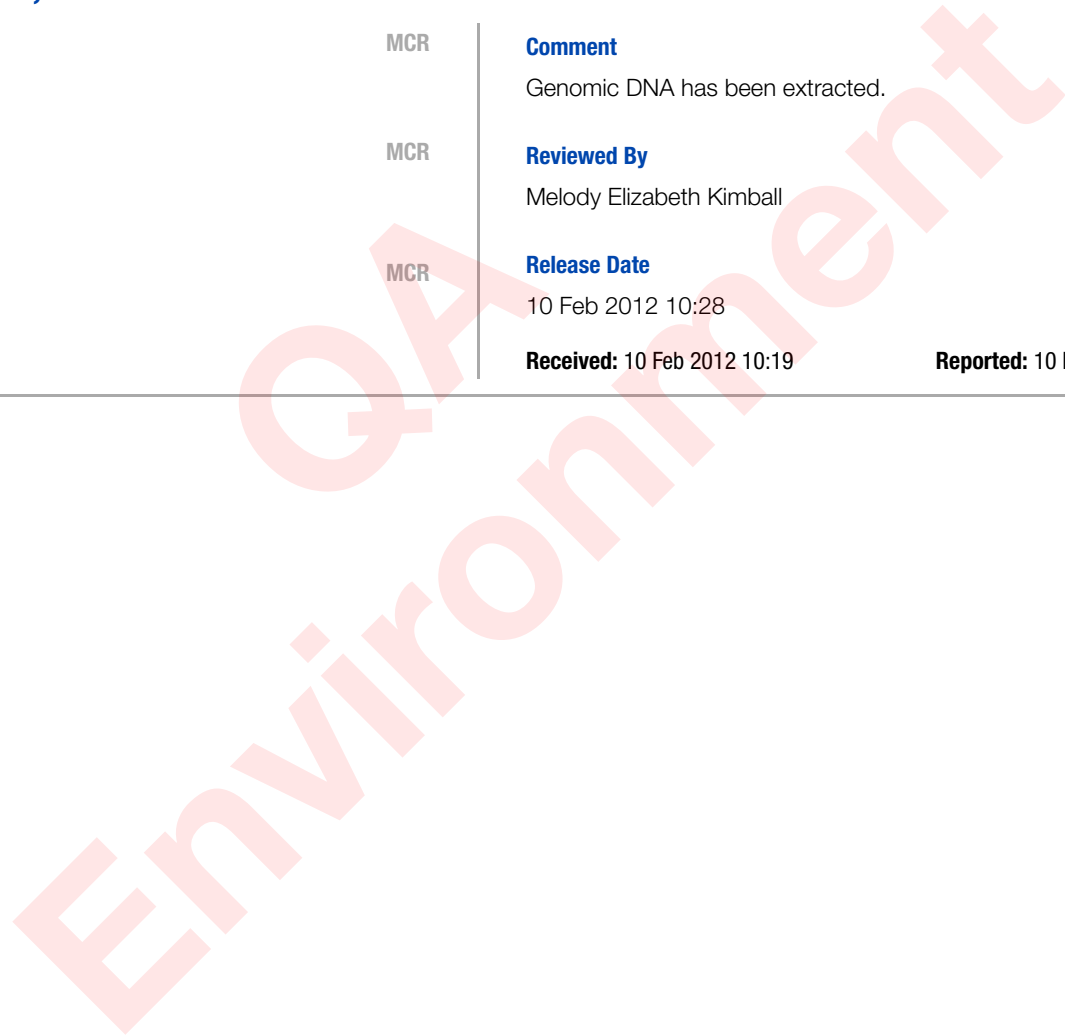
Release Date

10 Feb 2012 10:28

MCR

Received: 10 Feb 2012 10:19

Reported: 10 Feb 2012 10:31



Performing Site Legend

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



Patient ID SA00043484	Patient Name SAMPLEREPORT, PT1K	Birth Date 1966-06-10	Gender F	Age 45
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PTPN11 Gene, Known Mutation, B

PTPN11 Gene, Known Mutation

PTPN11 Known Mutation Result

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The PTPN11 Exon 3, nucleotide c.236A>G, amino acid p.Gln79Arg (p.G79R) familial mutation was not detected in this individual.

PTPN11 Known Mut Interpretation

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The PTPN11 p.Gln79Arg (p.G79R) mutation was previously identified in a family member with features of Noonan syndrome. Since this mutation 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 PTPN11 gene (GenBank accession number NM_002834.3), 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

Jamie Bruflat

PTPN11, Known Mutation Sequencing

MCR

Performed

Received: 10 Feb 2012 10:19

Reported: 10 Feb 2012 10:24

Performing Site Legend

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