

Patient Name SAMPLEREP,MAPTK A	Patient ID SA00057901	Age 46	Gender F	Order # SA00057901
Ordering Phys CLIENT,CLIENT				DOB 06/10/1966
Client Order # SA00057901	Account Information			Report Notes
Collected 05/30/2013 00:00	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
Printed 06/03/2013 10:46				

Test	Flag	Results	Unit	Reference Value	Perform Site*
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MAPT Known Mutation

RECEIVED: 05/31/2013 09:41 REPORTED: 06/03/2013 09:56

Specimen	Blood	MCR
Specimen ID	1061995	MCR
Order Date	03 Jun 2013 08:21	MCR
Reason For Referral		MCR

Family history of frontotemporal dementia associated with Parkinsonism (FTDP) or without Parkinsonism (FTD). Analyze the MAPT (tau) gene for the presence of a mutation.

Method MCR

DNA sequence analysis was used to test for the presence of the p.N279K (c.1842T>G) mutation in exon 10 of the MAPT (tau) gene. Testing was performed for this specific mutation because it was previously identified in an affected family member of this individual. Mutation nomenclature is based on the most common isoform (4R2N) (Human Mutation. 24(4):277-95, 2004 Oct.).

Result MCR

The following sequence change was detected:
Exon: 10
DNA change: c.1842T>G
Amino acid change: p.N279K (Asn279Lys)
Classification: DELETERIOUS

Interpretation MCR

This alteration is a known deleterious mutation.

This result is consistent with a diagnosis of frontotemporal dementia associated with Parkinsonism (FTDP) or without Parkinsonism (FTD) linked to a mutation in MAPT (tau) for this individual.

Since a mutation has been identified, testing of at risk family members is possible. Mutation-specific testing for MAPT is available at Mayo Medical Laboratories by ordering MAPTK/87925 MAPT Known Mutation. Please contact the Molecular Genetics Laboratory at 1-800-533-1710 with questions about this test. It is recommended that predictive testing is performed in conjunction with appropriate pre and post testing counseling.

A genetic consultation may be of benefit.

Unless reported or predicted to cause disease, alterations found deep in the intron or alterations that do not result

Performing Site Legend on Last Page of Report

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* Report times for Mayo performed tests are CST/CDT

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in an amino acid substitution are not reported. These and common polymorphisms identified for this patient are available upon request.

CAUTIONS:

Test results should be interpreted in context of clinical findings, family history, and other laboratory data. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.

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.

Bone marrow transplants from allogenic donors will interfere with testing. Call Mayo Medical Laboratories for instructions for testing patients who have received a bone marrow transplant.

Laboratory developed test.

Reviewed By:
Release Date

Jessica Rose Chavey
03 Jun 2013 09:54

MCR
MCR

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director:
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