MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
TESTING,87925				W2898060
Ordering Phys		**DOB**		
Client Order # W2898060	**Account Information** C7999998-STUSTEST 200 FIRST STREET SW ROCHESTER, MN 55901 (507)266-5730	**Report Notes**		
Collected 10/12/2009 14:02				
Printed 10/12/2009 17:46				

Test	Flag	Results	Unit	Reference Value	Perform Site*
MAPT Known Mutation			REPORTED 10/12/2009 14:24		
Specimen		DNA			MCR
Specimen ID		731624			MCR
Order Date		30 Jun 2009 11:39			MCR
Reason For Referral					MCR

Testing performed to confirm the presence of an alteration identified through research.

Method MCR

DNA sequence analysis was used to test for the presence of the A152T mutation in exon 7 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: 7
DNA change: 1405G>A (GCC>ACC)
Amino acid change: A152T (Ala152Thr)

Interpretation MCR

The significance of the A152T alteration is uncertain. This alteration has not been previously reported and therefore, we are unable to determine if this is a polymorphism or a disease-causing mutation.

This result does not rule out the presence of another alteration that may be responsible for the diagnosis of frontotemporal dementia. Disease-causing mutations may be present in other regions of the MAPT (tau) gene or in other genes. Reports in the literature suggest that mutations in the MAPT (tau) gene account for up to 40% of familial frontotemporal dementia, depending on the stringency of the clinical criteria used to establish the diagnosis.

Phenotypic overlap exists between FTDP-17 and FTD-PGRN. Therefore, genetic testing of the Progranulin (PGRN) gene (associated with FTD-PGRN) may be considered in this case. Please contact the molecular genetics laboratory at 1-800-533-1710 with questions about PGRN testing.

A genetic consultation may be of benefit.

Performing Site Legend on Last Page of Report

* Report times for Mayo performed tests are CST/CDT

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A list of common polymorphisms identified for this patient is 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.					
Reviewed By:		Dennis J O'Kane PhD			MCR
Release Date		08 Jul 2009 16:24			MCR

* Performing Site:

MCR	Mayo Clinic Dpt of Lab Med & Pathology 200 First St SW Rochester, MN 55905	Lab Director: Franklin R. Cockerill, III, M.D.

Patient Name	Collection Date and Time	Report Status
TESTING,87925	10/12/2009 14:02	Final
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