

#3336125
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UNSON-NEURO Research Fax:702

Apr 28 2009 08:41am P020/025

Quest Diagnostics Incorporated

LABORATORY RESULTS

Facsimile Copy

4230 Burnham Avenue, Suite 250
Las Vegas, NV 89119 (702) 733-7888

PATIENT ERICSON, CARL D

AUTORT2

11470

REFERRED BY UNIVERSITY NEUROLOGY ASSOC

AGE/SEX 11/06/61 M

ACCESSION# 49981645

1707 W. CHARLESTON STE. 220

COLLECTED 10/10/2007 14:34

MED. RECORD# 6205035103

LAS VEGAS, NV 89106

RECEIVED 10/10/2007 22:18

CHART# 451315657

FE0489 VENKATESH YEDATORE

TEST	RESULTS	REF RANGE	UNITS	LOW	NORM	HIGH
Patient Phone # (702)448-1858						
Ordering Physician: VENKATESH, YEDATORE S						
Collected 10/12/07 08:50AM						
DYSTONIA DNA [GN]	RESULT:					
Interpretation						
This individual does not possess the GAG946 deletion mutation in the DYT1 gene commonly associated with familial early onset generalized torsion dystonia and some cases of limb-onset focal dystonia.						
DYT1 allele 1: no mutation detected						
DYT1 allele 2: no mutation detected						
Methods						
Direct testing for the 3 base pair (GAG) deletion mutation (GAG946) in the DYT1 gene was performed by PCR amplification of genomic DNA followed by electrophoretic separation and size determination. This analysis, as performed here, is greater than 99% accurate.						
This test is performed pursuant to a PCR license agreement with Roche Molecular Systems, Inc.						
Comments						

THIS analysis identified two DYT1 alleles without the GAG946 mutation.

The GAG946 mutation is found in greater than 90% of familial early onset generalized torsion dystonia in those of Ashkenazi Jewish ancestry and at least 80% of early onset generalized torsion dystonia irrespective of ancestry. [1,2] The GAG946 mutation has also been identified, to date, in a small number of cases of limb-onset focal dystonia irrespective of ancestry and age of onset. [2]

References

1. Ozelius, L.J., et al. (1992) Am J Hum Genet 50: 619-628
2. Ozelius, L.J., et al. (1997) Nature Genetics 17: 40-48
(* = out of range)

GLUTAMIC ACID DECARBOXYLASE RESULT: *
[NI]

	RESULT	UNITS	REF RANGE
GAD-65 Ab			
GAD-65 AB	* 2.4	U/mL	1.0 OR LESS
	(* = out of range)		

CERULOPLASMIN	34		18-36	mg/dL	X
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COPPER, SERUM [NI] RESULT:

	RESULT	UNITS	REF RANGE
Copper			
Copper, Serum	148	mcg/dL	70-155

✓
10/31/02

Flu 9/2