Mount Sinai

Mount Sinai Genetic Testing Laboratory: ***Porphyria DNA Testing***

Department of Genetics & Genomic Sciences
Mount Sinai School of Medicine
One Gustave L. Levy Place, Box 1498
New York, NY 10029

Tel.: (212) 659-6783
Fax.: (212) 360-1809

Diagnostic Testing for Porphyria

Patient Information:

Name: **DUFF, STEPHEN B.**

Address: 105 S. Grand Avenue
Stratford, NJ 08084

Phone: 856-309-3739

DOB/Sex: 4/30/1952 / male

Lab. No.: P2008-167

Referring Physician:

Name: Dr. Rhona Schnur

Address: Cooper University Hospital
3 Cooper Plaza, Ste. #309
Camden, NJ 08103

Phone: 856-342-2069

Fax : 856-968-8315

Date Received: 8/15/2008

Sample: EDTA Blood

Requested Assay:

Mutation analyses of hydroxymethylbilane-synthase (HMBS), coproporphyrinogen oxidase (CPOX) and protoporphyrinogen oxidase (PPOX), the genes involved in the acute porphyrias, acute intermittent porphyria (AIP), hereditary coproporphyria (HCP) and variegate porphyria (VP), respectively.

Patient and Family History:

Stephen B. Duff is a 56 year old male with symptoms suggestive of an acute porphyria. Results of biochemical testing found slightly elevated urinary copro- and uroporphyrin, ALA, and PBG.

Results:

Mutation analyses by DNA sequencing did NOT identify a mutation in Stephen B. Duff's HMBS, CPOX and PPOX genes. All exons and intron/exon boundaries for each gene were sequenced.

Interpretation:

By sequencing genomic DNA, >97% of the known HMBS, >98% of the known CPOX and >98% of the known PPOX gene mutations listed in the Human Gene Mutation Database (www.hgmd.org as of 6/1/07) should be detected as well as most novel mutations. The absence of a specific mutation in Stephen B. Duff's HMBS, CPOX and PPOX genes significantly decreases the likelihood that Stephen B. Duff has AIP, HCP or VP. However, it is possible that the patient has a cryptic mutation in one of these genes which was not detected by sequencing.

Recommendations:

It is recommended that biochemical testing (measurement of urinary porphobilinogen (PBG) and aminolevulinic acid (ALA) and urinary and fecal porphyrins) be undertaken if the patient has symptoms of an acute porphyria attack, such as abdominal pain, peripheral neuropathy, and/or central nervous system involvement. Please contact us for referrals to certified biochemical laboratories that do specialized porphyria testing. Additional information about acute porphyrias can be obtained from the American Porphyria Foundation website (http://www.porphyriafoundation.com).

Genetic Counseling with regard to these test results is available through the Department of Genetics & Genomic Sciences, Mount Sinai School of Medicine, New York (1-212-659-6783

R. J. Desnick, Ph.D., M.D., Director

Date: 9/15/2008

FAXED dal 9/15/08