Genetic evaluation of AMPD1, CPT2, and PGYM metabolic enzymes in patients with chronic fatigue syndrome

P.E. Maltese[1], L. Venturini[2], E. Poplavskaya[3], M. Bertelli[1], S. Cecchin[1], M. Granato[1], S.Y. Nikulina[3], A. Salmina[3], N. Aksyutina[3], E. Capelli[4], G. Ricevuti[2] and L. Lorusso[5]

[1]MAGI Non-Profit Human Medical Genetics Institute, Rovereto, TN, Italy
[2]Cellular Pathophysiology and Clinical Immunology Laboratory, Department of Internal Medicine and Therapeutics, University of Pavia, Pavia, Italy
[3]Department of Internal Diseases N.1, Krasnoyarsk State Medical University, Krasnoyarsk, Russia
[4]Immunology and Genetic Analysis Laboratory, Department of Earth and Environmental Sciences, University of Pavia, Pavia, Italy
[5]Department of Neurology, Mellino Mellini Hospital, Chiari, BS, Italy

Corresponding author: P.E. Maltese
E-mail: paolo.maltese@assomagi.org

Genet. Mol. Res. 15 (3): gmr.15038717
Received April 19, 2016
Accepted July 22, 2016
Published July 29, 2016
DOI http://dx.doi.org/10.4238/gmr.15038717

ABSTRACT. Chronic fatigue syndrome (CFS) is a disease that can seriously impair one's quality of life; patients complain of excessive fatigue and myalgia following physical exertion. This disease may be associated with abnormalities in genes affecting exercise tolerance and physical performance. Adenosine monophosphate deaminase

(*AMPD1*), carnitine palmitoyltransferase II (*CPT2*), and the muscle isoform of glycogen phosphorylase (*PYGM*) genes provide instructions for producing enzymes that play major roles in energy production during work. The aim of this study was to look for evidence of genotype-associated excessive muscle fatigue. Three metabolic genes (*AMPD1*, *CPT2*, and *PYGM*) were therefore fully sequenced in 17 Italian patients with CFS. We examined polymorphisms known to alter the function of these metabolic genes, and compared their genotypic distributions in CFS patients and 50 healthy controls using chi-square tests and odds ratios. One-way analysis of variance with F-ratio was carried out to determine the associations between genotypes and disease severity using CF scores. No major genetic variations between patients and controls were found in the three genes studied, and we did not find any association between these genes and CFS. In conclusion, variations in *AMPD1*, *CPT2*, and *PGYM* genes are not associated with the onset, susceptibility, or severity of CFS.

Key words: *AMPD1*; *CPT2*; *PGYM*; Chronic fatigue syndrome; Polymorphism