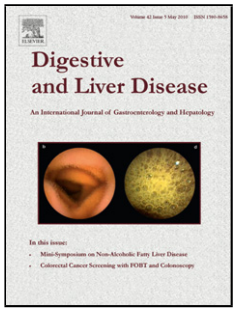
Accepted Manuscript



Title: Frequency of Thiopurine Methyltransferase mutation in patients of Mediterranean area with inflammatory bowel disease and autoimmune disorders

Author: Angela Di Salvo Carmelo Fabiano Vincenza Mannara Mariangela Dimarco Ambrogio Orlando Marco Affronti Fabio Salvatore Macaluso Mario Cottone

PII: S1590-8658(16)30703-4
DOI: http://dx.doi.org/doi:10.1016/j.dld.2016.08.125
Reference: YDLD 3249

To appear in: *Digestive and Liver Disease*

Received date: 10-5-2016
Accepted date: 5-8-2016

Please cite this article as: Di Salvo Angela, Fabiano Carmelo, Mannara Vincenza, Dimarco Mariangela, Orlando Ambrogio, Affronti Marco, Macaluso Fabio Salvatore, Cottone Mario.Frequency of Thiopurine Methyltransferase mutation in patients of Mediterranean area with inflammatory bowel disease and autoimmune disorders.*Digestive and Liver Disease* http://dx.doi.org/10.1016/j.dld.2016.08.125

TITLE: Frequency of Thiopurine Methyltransferase mutation in patients of Mediterranean area with inflammatory bowel disease and autoimmune disorders

AUTHORS: Angela Di Salvo[1], Carmelo Fabiano[2], Vincenza Mannara[1], Mariangela Dimarco[1], Ambrogio Orlando[1], Marco Affronti[1], Fabio Salvatore Macaluso[1], Mario Cottone[1].

INSTITUTIONS:

[1]Di.Bi.M.I.S., Division of Internal Medicine, "Villa Sofia-Cervello" Hospital, Via Trabucco 180, 90146 Palermo, Italy.

[2]Laboratory of Molecular Genetics, "Villa Sofia-Cervello" Hospital, Via Trabucco 180, 90146 Palermo, Italy.

CORRESPONDING AUTHOR: Mario Cottone, Professor of Internal Medicine; Division of Internal Medicine, ‘‘Villa Sofia-Cervello’’ Hospital, 90146 Palermo, Italy. Telephone: +39 0916802966. Fax: +39 0916802042. E-mail: mario.cottone@unipa.it

ABSTRACT

Background and aims: Few studies exist on the frequency of thiopurine methyltransferase (TPMT) mutation in patients from Southern Europe. We aimed to evaluate the frequency of TPMT mutation in a homogeneous Sicilian cohort of patients with inflammatory bowel disease (IBD), autoimmune and hematological disorders, the rate of thiopurine-related adverse events, and its association with the TPMT genotype.

Results: Among 105 patients with IBD, 45 with autoimmune disease, and 34 with hematologic diseases, the homozygous TPMT variant genotype was found in one patient only (0.5%), while the heterozygous TPMT genotype was identified in 8 patients (4.3%). In patients with IBD, leukopenia was observed in ten patients: one had the homozygous