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ABSTRACT BOOK

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THE ROLE OF POLYMORPHISM OF TIOPURINE METHYLTRANSFERASE IN THERAPY WITH AZATHIOPRINE. PRELIMINARY STUDY

F. Mortillaro^{1*}, M. Giammanco², F. Venturella¹

¹Department of Biological, Chemical and Pharmaceutical Sciences and Technologies, University of Palermo, Palermo, Italy; ²Department of Experimental Biomedicine and Clinical Neurosciences, University of Palermo, Palermo, Italy

*E-mail: francesca.mortillaro92@gmail.com

Azathioprine is an immunosuppressive medication used in the treatment of inflammatory diseases (MICI). Despite its extensive use in therapy, azathioprine can result in serious side effects such as myelosuppression. The likelihood of developing myelosuppression also depends on genetic factors: one of the enzymes involved in drug metabolism, methyltransferase tiopurine (TPMT), is subject to genomic polymorphism. To date, 40 polymorphisms have been identified, of which three are associated with a reduction in enzymatic activity. These are TPMT*2, TPMT*3A and TPMT*3C polymorphisms. TPMT*1 is the wild-type coding form for an enzyme capable of efficiently metabolizing the drug. The objective of the study was to determine the frequency of the three polymorphisms in a sample undergoing a TPMT genotype assay at the Molecular Laboratory of the "V. Cervello" hospital in Palermo. The test is carried out using DNA extraction procedures, PCRs, agarose gel electrophoresis and enzymatic digestion. From the analysis of 208 patients undergoing the test from 2011 to 2016, it was found that: 95.67% of patients did not have polymorphism; 2.40% had polymorphism *3A, 1.93% had polymorphism *3C. Therefore, 95.67% of patients may be exposed to azathioprine therapy without incurring side effects. Due to a reduced enzymatic activity, the remaining patients are able to be treated with azathioprine, provided that a lower dosage standard is used or is treated with another immunosuppressive drug.