Short communication

5-Fluorouracil toxicity-attributable IVS14 + 1G > A mutation of the dihydropyrimidine dehydrogenase gene in Polish colorectal cancer patients

Violetta Sulżyc-Bielicka¹*, Agnieszka Bińczak-Kuleta²*, Wiesława Pioch², Józef Kładny¹, Katarzyna Gziut³, Dariusz Bielicki³, Andrzej Ciechanowicz²

¹Department of Oncological Surgery, Pomeranian Medical University, Powstańców Wlkp. 72, PL 70-111 Szczecin, Poland
²Departments of Laboratory Diagnostics and Molecular Medicine, Pomeranian Medical University, Powstańców Wlkp. 72, PL 70-111 Szczecin, Poland
³Department of Gastroenterology, Pomeranian Medical University, Uniwersytecka 1, PL 71-252 Szczecin, Poland

*These authors contributed equally to this work

Correspondence: Andrzej Ciechanowicz, e-mail: aciech@sci.pam.szczecin.pl

Abstract:

DPYD gene encodes dihydropyrimidine dehydrogenase which is the initial and rate-limiting enzyme in the metabolism of 5-fluorouracil (5-FU). The aim of our study was PCR-RFLP based-genetic testing for the most common 5-FU toxicity-attributable IVS14 + 1G > A DPYD mutation (DPYD∗2A) in 252 Polish colorectal cancer (CRC) patients treated with this adjuvant chemotherapeutic regimen after surgery. The DPYD∗2A allele was identified only in one patient: a male who was one of 4 CRC patients suffering from grades 3–4 myelotoxicity upon 5-FU chemotherapy. We conclude that IVS14 + 1G > A DPYD (DPYD∗2A) variant occurs in the Polish population and is responsible for a significant proportion of life-threatening toxicity of 5-FU.

Key words:
dihydropyrimidine dehydrogenase, 5-fluorouracil toxicity, gene mutation