



Short communication

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

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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
