The R521K polymorphism of EGFR influences the risk of colorectal cancer

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In colorectal cancer (CRC) epidermal growth factor receptor (EGFR) family members (EGFR, HER2, HER3 and HER4) have been found frequently over-expressed. New therapies directed against EGFR have been developed in many human cancers. Unexpectedly, EGFR alterations could be good prognostic indicators, like in lung cancer, where an EGFR variant in non-smoker female patients is associated with higher survival after surgery and increases the efficiency of therapy based on EGFR inhibitors. The role of the genetic polymorphisms of the EGFR family members in colorectal cancer development has not been completely explored. In our preliminary study, three missense polymorphisms mapping in EGFR family members have been investigated in the peripheral blood of a small Italian sample size of 70 patients and 72 controls to verify if they could be considered CRC susceptibility factors. For the first time, the evidence of genotype association was found for the R521K EGFR polymorphism: the protective effect for this variant allele has been found to reduce the risk for colon cancer onset.

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