

GENETIC POLYMORPHISMS ASSOCIATED WITH COLORECTAL CANCER RISK

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BACKGROUND

Colorectal cancer (CRC) is currently the most frequent malignant gastrointestinal disease. Some recent publications have proposed that genetic polymorphisms (Single Nucleotide Polymorphism, SNP) in different genes may be potential markers of CRC risk.

PURPOSE

This study aimed to determine the association of SNP in *KIF9*, *PLCE1*, *MLH1*, *CYP2E1*, *TP53* and *SMAD7* genes with susceptibility to the development of CRC.

MATERIAL AND METHODS

A retrospective case-control study was performed, where 126 cases and 169 controls CRC of Caucasian ethnicity were included. The genotypes of the selected polymorphisms from *KIF9*, *PLCE1*, *MLH1*, *CYP2E1*, *TP53* and *SMAD7* genes were determined in different individuals using the real-time PCR with TaqMan probes. Then, the results were analyzed under different genetic models (additive, genotypic, allelic, dominant and recessive) to look for an association between them and CRC risk.

MLH1	rs1800734	G/A
KIF9	rs1076394	G/A
PLCE1	rs11187842	C/T
CYP2E1	rs1329149	C/T
TP53	rs1042522	G/C
SMAD7	rs4464148	T/C

RESULTS

G allele from SNP *MLH1* rs1800734 was found to be a protector marker for CRC in the genotypic model (OR_{AG vs AA}: 0,17; 95% CI: 0,05-0,49; p: 0,0015. OR_{GG vs AA}: 0,31; 95% CI: 0,10-0,80; p: 0,0217), beside gender and BMI in the multivariate statistical model, while the rest of polymorphisms were not found associated with CRC risk.

GENOTYPIC MODEL				
Variable	Ref (OR: 1,00)	Odds Ratio	95% CI	p-value
MLH1 rs1800734 AG	AA	0,17	0,05-0,49	0,00152
MLH1 rs1800734 GG	AA	0,31	0,10-0,80	0,02170
Gender	Mujer (♀)	1,82	1,06-3,15	0,03205
Obesity	No	0,58	0,34-1,00	0,04950

CONCLUSION

AA genotype from SNP *MLH1* rs1800734 is a marker of CRC risk.