

Supplementary Materials: Simultaneous Analysis of SEPT9 Promoter Methylation Status, Micronuclei Frequency, and Folate-Related Gene Polymorphisms: The Potential for a Novel Blood-Based Colorectal Cancer Biomarker

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Table S1. SNPs description according to HGVS recommendations §.

Gene-RefSNP	HGVS Nomenclature	DNA Variant and Protein	Reference
RFC-rs1051266	NC_000021.9:g.45537880T>C	c.80A>G [p.(His27Arg)]	RFLP * [48]
FOLR-rs2071010	NC_000011.10:g.72189920G>A	c.-20G>A	RT **
DHFR-rs70991108	NC_000005.10:g.80654344_80654345insTG GCGCGTCCCCGCCAGGT	c.86+59_86+60insACCTGG GCGGGACGCGCCA	RFLP * [49]
TS-rs45445694	NC_000018.10:g.647646_647673CCGCGCC ACTTGGCCTGCCTCCGTCCCG [2-4,7-9]	c.*34+169_*34+196CGGGACGGAGG CAGGCCAAGTGGCGCGG [2-4,7-9]	RFLP * [50]
SHMT-rs1979277	NC_000017.11:g.18328782G>A	c.1420C>T [p.(Leu474Phe)]	RT **
MTHFR-rs1801133	NC_000001.11:g.11796321G>A	c.665C>T [p.(Ala222Val)]	RT **
rs1801131	NC_000001.11:g.11794419T>G	c.1286A>C [p.(Glu429Ala)]	RT **
MTRR-rs1801394	NC_000005.10:g.7870860A>G	c.147A>G [p.(Ile49Met)]	RT **

§ HGVS nomenclature version 2.0, searched through Mutalyzer 2.0.beta-32, released on 26 June 2014;

* RFLP = PCR-RFLP analysis carried out according to published methods (reference parenthetically);

** RT = Real-Time PCR with TaqMan allelic discrimination assay (Applied Biosystem, Foster City, CA, USA).

Table S2. Allele frequency in CRC patients (cases) and controls.

Genotypes Distribution <i>n</i>					
Subjetcs	WT	HE	SNP	MAF *	<i>P</i> <i>HWE</i>
RFC-rs1051266					
Cases	8	12	4	<i>q</i> = 0.417	0.889
Controls	9	13	4	<i>q</i> = 0.404	0.845
FOLR-rs2071010					
Cases	23	1	0	<i>q</i> = 0.021	0.917
Controls	22	4	0	<i>q</i> = 0.077	0.671
DHFR-rs70991108					
Cases	13	9	2	<i>q</i> = 0.271	0.804
Controls	7	14	5	<i>q</i> = 0.461	0.671
TS-rs45445694					
Cases	6	16	2	<i>q</i> = 0.417	0.069
Controls	4	10	12	<i>q</i> = 0.654	0.443
SHMT-rs1979277					
Cases	16	7	1	<i>q</i> = 0.188	0.834
Controls	14	11	1	<i>q</i> = 0.250	0.513
MTHFR-rs1801133					
Cases	7	13	4	<i>q</i> = 0.437	0.622
Controls	6	14	6	<i>q</i> = 0.500	0.695
MTHFR-rs1801131					
Cases	15	8	1	<i>q</i> = 0.208	0.959
Controls	11	13	2	<i>q</i> = 0.327	0.488
MTRR-rs1801394					
Cases	13	9	2	<i>q</i> = 0.271	0.804
Controls	6	12	8	<i>q</i> = 0.538	0.716

* MAF: Minor allele frequency.