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Título	Single nucleotide variants c.-13G → C (rs17429833) and c.108C → T (rs72466472) in the CLDN1 gene and increased risk for familial colorectal câncer
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Assunto (palavras chaves)	CLDN1 gene; Familial colorectal câncer; Single nucleotide variants; SNVs c.-13G → C (rs17429833); c.108C → T (rs72466472); c.369T → C (rs9869263)c.370G → A (rs140846629); Susceptibility
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Resumo	Background: The Claudin-1 (CLDN1) protein plays an important role in the function of the tight junction and studies have shown it is aberrantly downregulated in many tumors including colorectal cancer (CRC). The aim of this study was to determine the relationship between four SNVs in the CLDN1 gene [c.-13G → C (rs17429833), c.108C → T (rs72466472), c.369T → C (rs9869263), and c.370G → A (rs140846629)] and the risk of familial colorectal cancer (FCC). Methods: A case-control study was conducted with peripheral blood DNAs from 50 patients with CRC that belong to FCC families and 96 healthy control individuals. The analysis of genetic variants was performed by PCR and restriction enzymatic digestion. Results: The patients and control groups presented in Hardy-Weinberg equilibrium for all evaluated SNVs. No significant differences occurred in wild-type homozygous, heterozygous and variant homozygous genotypes, separately or together, in patient and control groups for the SNVs rs72466472, rs9869263, and rs140846629. However, for the SNV rs17429833, increased frequency of GC genotype occurred in patients compared to healthy individuals (58.30% vs. 41.70%), with an OR = 3.28 (95%CI = 1.22 to 9.09) for CRC. In the patients' group, individuals harboring combined genotypes rs17429833 (GC) and rs72466472 (CC) (26% vs. 8.42%) showed an OR = 3.78 (95%CI = 1.33 to 11.48). Moreover, patients harboring GC genotype for SNV rs17429833 presented significantly association with well differentiated adenocarcinoma when compared to moderately differentiated adenocarcinoma [60% vs. 22.58%, OR = 6.3 (95%CI = 1.15 to 39.76)].

	Conclusions: The GC genotype for the SNV rs17429833 or combined genotypes for SNVs rs17429833 (GC) and rs72466472 (CC) seems to be risk factors for patients with FCC in Brazilian patients; however, a larger number of patients needs to be evaluated to confirm our results.
Fomento	