ASSOCIATION OF rs2234693 AND rs1643821 POLYMORPHISMS OF ESR1 GENE WITH RISK TO HEPATOCELULAR CARCINOMA

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Introduction and objectives: The hepatocellular carcinoma (HCC) is a primary tumor of the liver that represents the fifth most common malignancy of the world. Genetic variation has been discussed as one responsible for the variable risk of developing HCC, being the molecular polymorphisms, mainly the Single Nucleotide Polymorphisms (SNPs), the most studied. A several SNPs were described for ESR1 gene, including rs2234693 and rs1643821. Numerous studies highlighted the effect of sexual hormones in the function and morphology of liver, being important to study the influence of different polymorphisms of estrogen receptor in HCC. The objective of this study was to investigate the association between the polymorphisms cited to the risk of developing HCC and analyze some parameters as age, sex and etiology.

Material and methods: Samples of liver specimens from paraffin-embedded tissue of 51 patients with HCC and 55 patients control without the disease were collected from Laboratório KCM. It was realized the DNA extraction, quantification by spectrometry and analysis of the polymorphisms cited by PCR Real Time. It was calculated the allelic and genotypic frequency and applied the Chi-square test (p<0.05). Hardy-Weinberg balance tested.

Results and conclusions: Male gender was the most prevalent and the mean age was 53 years. The etiology found most often in cases was the association of cirrhosis with hepatitis C virus. A significant difference between cases and controls were only found for the SNP rs1643821. The rs1643821 SNP seems to be associated with some influence in promoting the development of HCC.

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