Genetic Markers of Predisposition to Thyroid Cancer: A Pilot Study in Rio Grande do Norte

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Introduction: Thyroid cancer represents about 1% of all malignancies, and its incidence has increased substantially worldwide in the last three decades. Genetic predisposition to this cancer might be associated with multiple low to moderate penetrance genes that interact with each other and with the environment. However, susceptibility genes for this disease are not well known. **Objectives:** We aimed to verify whether the main single-nucleotide polymorphisms (SNPs) associated with predisposition to thyroid cancer are present in a Brazilian population. **Materials and Methods:** Ninety formalin-fixed paraffin-embedded tissue samples from patients with thyroid cancer were obtained from Hospital Liga Norte Riograndense Contra o Câncer archives. Samples were genotyped and compared with 180 genotypes available in the HapMap database, of which 90 were from European and 90 from African populations, respectively. Samples from case group were genotyped using the MALDI-TOF based Mass Array genotype platform and iPlex primer extension (Sequenom). Statistical analyses were performed using SNPassoc function within the R statistical package. **Results and Discussion**: Eleven polymorphisms were excluded from the analysis, because they were not in Hardy-Weinberg equilibrium ($p<0.05$). When comparing case and control groups, eight variants were significantly associated with an increased risk of developing thyroid cancer: rs3744962, rs258107, rs4075022, rs9943744, rs4075570, rs17485896, rs944289 and rs2651339 (OR >1.0). On the other hand, three SNPs showed a protective effect: rs2356508, rs99931 and rs1461855 (OR <1.0). Finally, polymorphisms rs374492 C/T, rs2588107 C/T and rs4075022 C/T were associated with a relative risk of 3.78 ($p= 0.0000627$), 2.91 ($p= 0.00008272$) and 2.35 ($p= 0.002011$), respectively. **Conclusion:** SNPs rs3744962, rs258107, rs4075022, rs9943744, rs4075570, rs17485896, rs944289 and rs2651339 might be potential markers of predisposition to thyroid cancer in Rio Grande do Norte population. However, other studies with a control group constituted of samples from the same region as the case group should be conducted to confirm these results.

Keywords: Thyroid Cancer, Polymorphism, Genetic Predisposition.