DOI: 10.29289/259453942019V29S1CP05

FIRST STUDY OF THE PREVALENCE OF THE TP53 P.R337H MUTATION IN BREAST CANCER PATIENTS IN THE CENTRAL REGION OF BRAZIL

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Breast cancer is the second most common type of cancer in the world and the most common among women. Of the total number of cancer cases diagnosed each year, it is estimated that 5 to 10% are hereditary, usually caused by mutations in tumor suppressor genes. Mutations in the TP53 gene are the most common genetic alterations in human malignant tumors and are primarily responsible for Li-Fraumeni Syndrome. Several cancers may be related to this syndrome and the most common is breast cancer. The mutation of the TP53 gene p.R337H has a high prevalence in the South and Southeast regions of Brazil, where its presence was observed in about 0.3% of the population in general. Despite the high incidence of this pathogenic mutation in these regions of Brazil, it is practically not known what the prevalence in the Midwest region is. Therefore, the present study had the initiative to describe, for the first time, the prevalence of the pathogenic mutation p.R337H of the TP53 gene in patients with breast cancer in the central region of Brazil. Among the 102 patients analyzed, a woman with the p.R337H mutation in heterozygosis was identified. These results suggest that the prevalence of this mutation in the Midwest is 1% (1/102) among breast cancer patients, a lower prevalence compared to studies in the South and Southeast regions that identified this variant in 2.5% and 8.5% of patients with breast cancer. We suggest that this mutation may be less frequent in the Midwest than in other regions of Brazil. This study reinforces the hypothesis that the founding haplotype of the p.R337H mutation appeared in the South region and moved through the Southeast, which justifies the high incidence of this mutation in these regions.