DOI: 10.29289/259453942018V28S1023

ANALYSIS OF POLYMORPHISMS IN THE TP53 GENE IN PATIENTS WITH CLINICAL DIAGNOSIS FOR HEREDITARY BREAST CANCER

Paula F. F. Silva[1], Rebeca M. Goveia[1], Thais B. Teixeira[2], Hugo Deleon[1], Gabrielly R. Abrantes[1], Kézia A. Delmond[1], Aliny P. de Lima[1], Jeffter R. Silva[1], Elisângela de P. S. Lacerda[1]

[1]Laboratory of Molecular Genetics and Cytogenetics, Institute of Biological Sciences I (ICB I), Universidade Federal de Goiás – Goiânia (GO), Brazil.
[2]Hospital das Clínicas, Universidade Federal de Goiás – Goiânia (GO), Brazil.

Breast cancer is the second most frequent type of cancer in the world and the most common among women. Of the total number of cancer diagnosis each year, it is estimated that 5% to 10% are hereditary, usually caused by mutations in tumor suppressor genes. Mutations or polymorphisms in the TP53 gene are the most common genetic alterations in human malignant tumors. More than 85 polymorphisms in the TP53 gene have been identified, with PIN2 located in exon 2, PIN3 located in exon 3 (16pb doubling) and PEX4 located in exon 4 (Arg72Pro). Studies have reported that PIN2 has no pathogenic significance, however PEX4 and PIN3 have inconclusive results, and there may be some association between the risk of developing cancer and the presence of these variants. The objective of this work was identify the polymorphisms present in exons 2 to 4 of the TP53 gene in patients with clinical diagnosis for hereditary breast cancer. For the study, 5 mL of blood were collected from 55 female patients treated at the Hospital das Clínicas of the Federal University of Goiás. After collection, the samples were submitted to DNA extraction, Polymerase Chain Reaction (PCR) and sequencing Sanger of exons 2, 3 and 4 of the TP53 gene. The results showed that three polymorphisms were found: 31 (56.3%) had the PIN2 polymorphism, 20 (36.3%) had PEX4, and only one (1.8%) presented PIN3, some sequences presented more than one polymorphism, only 16 (29.0%) did not present any polymorphism in the analyzed regions. In addition to these three polymorphisms, an unknown polymorphism was verified in exon 3, which is not present in the databases nor in the literature, suggesting that it may be a polymorphism not yet described until now. Patients' charts were also analyzed and no association between polymorphisms and age at diagnosis, treatment response, and patient survival were identified. These results contribute to the identification of polymorphisms present in the TP53 gene and show the relatively high frequency among breast cancer patients. However, further studies are needed to better understand the influence of these polymorphisms on the TP53 gene and the risk of developing hereditary breast cancer.