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Variant analysis in the TP53 gene for families in the state of Goiás with suspected Li-Fraumeni syndrome: tool for early diagnosis and prevention of breast cancer

Késsila Macedo Veiga[1], Rebeca Mota Goveia[1], Paula Francinete Faustino da Silva[1], Adelaide Fernandes Costa[1], Elisangela de Paula Silveira Lacerda[1]

[1]Universidade Federal de Goiás, Institute of Biological Sciences, Center for Human Genetics.

Objective: Breast cancer is the most common type of cancer in the world. In Brazil, there is a high incidence of Li-Fraumeni syndrome (LFS), one of the main syndromes related to the development of hereditary breast cancer. This study aimed to identify the prevalence of variants in the TP53 gene in probands suspected of having LFS and their families. **Methodology:** We evaluated 123 patients who met the National Comprehensive Cancer Networking criteria for suspected hereditary breast cancer syndromes, who were referred to the Center for Human Genetics/Universidade Federal de Goiás (UFG) by Hospital das Clínicas/UFG. After applying the Informed Consent Form, 4 mL of venous blood was collected for DNA extraction used for next-generation sequencing and analysis of the entire coding region of the TP53 gene. The DNA library was prepared using the PCR target amplification method with the Oncomine™ BRCA Expanded panel kit and subjected to sequencing on the Ion Torrent platform. Raw data were evaluated on the Ion Reporter platform, and variants were classified according to the American College of Medical Genetics. **Results:** Of the 123 patients evaluated, 12.19% (15/123) were positive for TP53 variants in seven different families. Variants c.1010 G>A (6/7) and c.455C>T (1/7) were identified. Within the families with variants, it was analyzed that 47.05% (8/17) of the family members tested were also positive cases, with 100% (17/17) not developing any type of cancer to date. **Conclusion:** These data alert to the considerable incidence of LFS in the state of Goiás and draw attention to the power of directing treatment and prevention of breast cancer that genetic tests could provide for our population, with the possibility of personalization monitoring high-risk families.

Keywords: hereditary breast cancer; NGS; Li-Fraumeni syndrome; TP53.