https://doi.org/10.29289/259453942021V31S2003

R337H MUTATION AND DUCTAL CARCINOMA IN SITU OF THE BREAST

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Objective: To identify the prevalence of the founding variant c.1010G>A (R337H) in the TP53 gene in patients with ductal carcinoma in situ (DCIS). **Methods:** This is a cross-sectional study, carried out using data from the Goiania Population-Based Cancer Registry, in partnership with the team from the Molecular and Cytogenetic Genetics Laboratory of the Federal University of Goias and Clinical Research Unit/HC. Peripheral venous blood collections, DNA extraction, and Sanger genetic sequencing were carried out in 27 samples from unselected patients diagnosed with DCIS in the period between 1994 and 2010, who agreed to participate in the research by signing the free and informed consent form. This work was approved by the Research Ethics Committee of Hospital das Clínicas/UFG, as a proposing institution, and Research Ethics Committee of the Co-participant Institution Hospital Araújo Jorge/Association for Combating Cancer in Goiás (ACCG) in attention to the resolution CNS: 466/2012 and its complementaries (1,940,921). **Results:** In our study, conducted in mid-western Brazil with a population of women with DCIS not selected for family history and involving 27 samples, 2 cases (7.4%) of the pathogenic TP53 R337H mutation were found. **Conclusions:** The present study showed that the prevalence of the founding variant c.1010G>A (R337H) in the TP53 gene in patients with DCIS proved to be considerably high, comparing the same rate found in other Brazilian studies for invasive breast carcinoma. This study warrants that there is a need for further studies testing not only the TP53 gene, but also other genes related to hereditary breast and ovarian cancer syndrome in DCIS. Although DCIS is a non-obligate precursor to invasive carcinoma, we highlighted the facts that strengthen the reflection on better genetic research in women with DCIS.

Keywords: Epidemiology; Carcinoma; Intraductal; Noninfiltrating; Survival; Breast Neoplasms; Hereditary Breast and Ovarian Cancer Syndrome; Li–Fraumeni Syndrome.