

GENETICS

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PREVALENCE OF VARIANTS OF UNCERTAIN SIGNIFICANCE IN TESTS REQUESTED FOR BREAST CANCER PATIENTS IN A PRIVATE SERVICE

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Introduction: The genetic mutations test among breast cancer (BC) patients is one of the steps for the diagnosis in the majority of the patients. To identify and manage patients with hereditary predisposition to cancer is also a competence of the breast surgeon. The development of Next Generation Sequence (NGS) has allowed the reduction of the tests' cost as well as the expansion of the analyzed genes, besides BRCA 1 and 2, and the inclusion of new genes of high and moderate penetrance. There is a concern about the impact of these results because there is not a well-established conduct for all the mutations as well as for the increase of the diagnoses of variant of uncertain significance (VUS) diagnosed in the panels, mostly in patients that did not receive a formal genetic counseling. Studies show that the larger number of analyzed genes is related to a better chance of detecting VUS, reaching 40%, but they are not conduct modifiers. The literature shows that approximately 90% of VUS are reclassified as benign. **Objectives:** To assess prevalence of VUS in multigenic panels requested by the non-geneticist physician, in private office, performed on patients with BC diagnosis. **Methods:** A retrospective cross-sectional study was conducted based in data from invasive BC patients or in situ or with high risk for neoplasia that attended a private office and were subjected to multigenic panels requested by the non-geneticist physician from January 2019 to January 2020. Statistical analysis frequency measurements were analyzed in Excel Office®. **Results:** 147 patients underwent the genetic test of 83 genes with NGS technology. Only one was a male. Among the tests performed, 48 were negative for pathogenic variants and 23 were positive for pathogenic mutations in 22 (15%) patients, the most common being in BRCA2 gene (7 cases), followed by MUTHY (6 cases). 137 VUS occurred in 77 (52.4%) patients, the most common of these being in gene POLE and RECQL4. **Conclusions:** The data found in our population match the literature, showing more than half of the patients with VUS. This demonstrates the importance of test interpretation as well as inpatient correct orientation.