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# 1196 – Discovery of a novel variant in the BRCA2 gene related to breast cancer in a patient with early diagnosis

Manuela Rocha Matos Rezende<sup>1</sup>, Paula Francinete Faustino Silva<sup>1</sup>, Flávia Barreto Sousa<sup>1</sup>, Elisangela Paula Silveira-Lacerda<sup>1</sup>

<sup>1</sup>Universidade Federal de Goiás – Goiânia (GO), Brazil.

**Introduction:** Hereditary breast cancer is frequently associated with pathogenic variants in the BRCA1 and BRCA2 genes, and the identification of new alterations is essential for advancing clinical oncogenetics and genetic counseling.

**Objective:** To report the identification of a novel variant in the BRCA2 gene in a patient with early-onset breast cancer and to discuss its possible clinical relevance.

**Methods:** A 31-year-old woman diagnosed with invasive ductal breast carcinoma was referred for oncogenetic evaluation. DNA was extracted from peripheral blood, followed by analysis using next-generation sequencing, including the genes BRCA1, BRCA2, ATM, BARD1, BRIP1, CHEK2, PALB2, and TP53. Variant interpretation followed the criteria established by the American College of Medical Genetics and Genomics/Association for Molecular Pathology (ACMG/AMP, 2015), supported by public and clinical databases. Pre-test genetic counseling addressed indications and limitations, while post-test counseling provided proper interpretation of results and family guidance.

**Results:** A nonsense variant in the BRCA2 gene (c.8786delT) was identified, resulting in a premature stop codon (p.Leu2929Ter) with a likely loss of protein function. The variant was not found in databases such as gnomAD, ClinVar, dbSNP, and ABRAOM, suggesting a rare and possibly novel variant. According to the ACMG/AMP criteria, it was classified as likely pathogenic.

**Discussion:** The identification of this variant reinforces the importance of genetic sequencing, especially in underrepresented populations, and contributes to the understanding of genetic variability associated with breast cancer. It also highlights the relevance of integrating these findings into clinical practice, thereby enabling more accurate diagnosis, prevention, and management, as well as strengthening national databases.

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**Keywords:** hereditary breast cancer; BRCA2; novel variant; oncogenetics; NGS.