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EARLY-ONSET BREAST CANCER PATIENTS FULFILLING HEREDITARY BREAST AND OVARY CANCER AND LI-FRAUMENI-LIKE SYNDROMES CAN HARBOR TP53 PATHOGENIC VARIANTS

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Objective: We investigate the prevalence of TP53 germline pathogenic variants in a cohort of 83 breast cancer patients and 217 family members from the Midwest Brazilian region. **Methods:** All patients met the clinical criteria for hereditary breast and ovarian cancer syndrome (HBOC) and were negative for BRCA1 and BRCA2 mutations. Moreover, 40 index patients fulfilled HBOC and the Li-Fraumeni-like syndromes (LFL) criteria. The samples were tested using next-generation sequencing for TP53. **Results:** Three patients harbored TP53 missense pathogenic variants (p.Arg248Gln, p.Arg337His, and p.Arg337Cys), confirmed by Sanger sequencing. One patient showed a large TP53 deletion (exons 2–11), which was also confirmed. The p.R337H variant was detected in only one patient. **Conclusion:** This study concluded that 4 out of 83 HBOC and LFL patients presented TP53 pathogenic variants at a young age. In contrast to other Brazilian regions, the TP53 p.R337H variant appeared with low prevalence.

Keywords: TP53. Breast cancer. Li-Fraumeni syndrome. Cancer predisposition.