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SOMATIC MUTATIONAL LANDSCAPE CHARACTERIZATION OF METASTATIC BREAST CANCER IN BRAZIL

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Objective: Breast cancer (BC) is the most common malignancy among Brazilian women after non-melanoma skin cancer. The mutational landscape of BC in Brazil is unknown. This study describes the mutational profile of a cohort of patients with metastatic breast cancer (MBC) who had undergone next-generation sequencing (NGS) using a comprehensive somatic tumor panel. **Methods:** We retrospectively reviewed medical records from MBC patients. The mutational profile, clinical, and demographic characteristics were abstracted. Furthermore, the patterns of ordering the panel and its usefulness for a clinical decision were evaluated. **Results:** We found 54 female patients who fulfilled the above criteria. The median age was 58 years (32–86). Most tumors tested were hormone receptor-positive (74%), followed by triple-negative (20.3%), hormone receptor-positive/HER2-positive (3.7%), and HER-2 positive (1.85%). The median time between the diagnosis of metastatic disease and the NGS execution was 40 months (0–112), and only three patients (5.5%) had not received systemic treatment prior to the test recommendation. Somatic mutations were identified in 94.4% (n=51) of the patients, mainly in PIK3CA (48.1%), TP53 (42.5%), and ESR1 (18.5%) genes. Tumor burden mutation (TMB) was informed in 61.1% (n=33) somatic panels, and 15.1% (n=5) had tumors with TMB ≥ 10 mutations/megabase. Approved genome-driven cancer therapy was found in 54.9% (n=28), and eight patients (28.5%) received it. **Conclusion:** This study showed a high proportion of actionable somatic genomic alterations, and it reinforces the growing usefulness of a comprehensive NGS tumor somatic panel in managing patients with MBC.

Keywords: Breast cancer. Next-generation sequencing.