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### Characteristics of Breast Cancer in Carriers of Pathogenic Variant in ATM in a Large Academic Health Center

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**Characteristics of breast cancer in carriers of pathogenic variant in ATM in a large academic health center**

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Background:

Pathogenic variants (PVs) in ATM are associated with an increased risk of breast, pancreas, and other cancers. The clinical and pathological characteristics of ATM-associated breast cancers have not been well defined.

Methods:

Patients who underwent multigene panel testing between December 2012 and May 2023 and were identified to harbor ATM PVs were included in the study. We analyzed demographics, germline findings, histopathology, genomic recurrence score, and management of ATM PV carriers with breast cancer.

Results:

A total of 222 individuals were identified to have PVs in ATM, of whom 184 (83%) were female. The majority were Caucasian (n = 179, 81%) and of those, 22 (12%) had Ashkenazi Jewish ancestry. The median age at genetic testing was 52y, with a range of 20-90y. A total of 111 patients (50%) had a personal cancer diagnosis, the most common of which was breast cancer (n = 70; 63%). The median age at breast cancer diagnosis was 53y, with a range of 25-84y. The most common histopathology was invasive ductal carcinoma (74%), followed by DCIS (21%), and invasive lobular carcinoma (5%). The majority of patients had breast tumors with stage 2 or 3 (78%), grade 2 or 3 (92%), ER and/or PR positive (92%) and HER2 negative (70%). One patient had triple-negative breast cancer. Forty-four percent of breast cancers were less than two centimeters, 23% were lymph node-positive, and 29% had lymphovascular invasion. Thirty-eight percent of patients underwent bilateral mastectomy, 54% received radiation therapy, 40% received adjuvant chemotherapy, 24% received neoadjuvant chemotherapy, and 70% received hormonal therapy. Of the 15 breast cancers with known 21-gene recurrence scores, 47% were intermediate (11-26), 33% were high ( $\geq 26$ ) and 20% were low ( $\leq 10$ ). Other notable cancers observed were pancreas (n = 9), colorectal (n = 6), prostate (n = 5), uterine (n = 4), melanoma (n = 4), and thyroid (n = 4).

Conclusion:

Our study outlines the clinical and pathological characteristics of ATM-associated breast cancer in a large academic center. The majority of breast cancers were invasive ductal type, early stage, intermediate or high grade, and hormone receptor-positive with an intermediate or high recurrence score. Despite the early stage, a significant proportion of patients underwent bilateral mastectomy. Further studies are needed to better elucidate the unique characteristics of breast cancer in ATM PV carriers in order to provide tailored management guidelines for this population.