

The Clinical Utility of Genetic Testing for Moderate Penetrant Breast Cancer Genes: A Systematic Review of Cases

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ATM, *CHEK2*, *NBN* and *PALB2* are moderate risk cancer genes that confer a 17-58% lifetime risk of breast cancer. This study describes the clinical utility of testing these genes in families at elevated risk of breast cancer. Patients in this study carry pathogenic mutations in *ATM*, *CHEK2*, *NBN*, or *PALB2*. Each pedigree was analyzed to determine what breast cancer risk management options would be recommended based upon personal and family cancer history data alone. This was compared to the recommendation given to the patient following the identification of a pathogenic mutation. Probands were contacted to report what surgical or screening options they pursued and what communication and cascade testing has occurred within their families. 81.0% of women affected by breast cancer and 47.1% of unaffected women gained access to annual breast MRI due to their mutation. 19% of affected women and 52.9% of unaffected women would have been recommended annual breast MRI based on personal and family history data alone. 29.7% of affected women underwent risk reducing mastectomy and 17.6% of unaffected women underwent prophylactic mastectomy. 69.8% of probands informed all first-degree relatives of their genetic test result, and 22.9% of first-degree relatives pursued genetic testing. 71.7% of first-degree female relatives could increase their breast cancer screening should they test positive for the familial mutation. Genetic testing for moderate risk breast cancer genes offers clinical utility because probands and their family members gain access to risk management options that would not be available to them otherwise.