Increasing evidence supports the benefit of identifying BRCA1 and BRCA2 germline mutations in early breast cancer. Selection of patients for genetic testing is based on defined criteria taking individual and family history related factors into account. It is important to make a distinction between efficacy and effectiveness of BRCA testing criteria. Efficacy can be defined as the performance under ideal circumstances, whereas effectiveness refers to its real life performance. To allow for an unbiased and detailed evaluation of efficacy and effectiveness of the Swedish BRCA testing criteria, we retrospectively analyzed a prospectively collected cohort of 273 breast cancer patients from the well-characterized, population-based, single-site All Breast Cancer in Malmö (ABiM) study. The patients were diagnosed with breast cancer during the years 2007 through 2009. Out of 20 mutation carriers identified, 13 fulfilled Swedish criteria at time of diagnosis. Thus, the efficacy of these criteria was 65%. Excluding three patients in whom a mutation was already known at time of diagnosis, only 3/17 had been identified in the clinical routine, corresponding to an effectiveness of 18%. Here we detail the reasons why mutation carriers in our cohort were not detected though routine health care. In conclusion, effectiveness of BRCA testing criteria was much lower.
than efficacy. Our results indicate that current testing criteria and procedures associated with BRCA1 and BRCA2 testing are insufficient. There is room for improvement of their efficacy, but even more so regarding effectiveness. Clinical BRCA testing routines need to be critically revised.

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