

Registry Case Study: Universal Breast Cancer Registry

A biopharma partner wished to increase use of their multigene panel vs single-gene testing through NCCN Guidelines expansion



OUR APPROACH

- > Conduct an observational disease registry enrolling 1,000 patients with breast cancer at 25 Axess member sites
- > Survey sites to ensure enrollment at a 1:1 ratio between patients meeting NCCN Guidelines criteria for hereditary cancer genetic testing and those who do not
- > Implement sponsor's panel test across all participating sites



RESULTS

- > Restricting 80-gene panel testing to only those patients who meet NCCN Guidelines criteria would miss many carriers, as no significant difference was found between those who met the criteria and those who did not



IMPACT

- > The American Society of Breast Surgeons amended their genetic testing guidelines on the basis of the registry results
- > Payor groups increased funding for genetic testing, citing the revised guidelines

THE AMERICAN SOCIETY OF Breast Surgeons - Official Statement -
Consensus Guideline on Genetic Testing for Hereditary Breast Cancer

Underdiagnosis of Hereditary Breast Cancer: A Genetic Testing Guidelines a Tool or an Obstacle?
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Hereditary Cancer Risk: A Growing Body of Evidence Supporting Broader Testing

Background	Results	Results NCCN Guidelines	Panel Size and Test Outcome	Conclusions
<p>Among 221 breast cancer patients, 11.8% had a positive result for a pathogenic mutation. The most common positive findings were in BRCA1, BRCA2, CHEK2 and MLH1.</p>	<p>Among patients who met NCCN criteria for testing, 13.8% had pathogenic mutations (10.7% of which were in BRCA1/2), whereas 11.7% of the patients who did not meet NCCN criteria did. Pathogenic test pathogenic mutations (28.0% of total) were in BRCA1/2.</p>	<p>Patients under 50 had pathogenic mutations: 15.1% (10/66) patients over 50, 12.1% (10/82) of the total. A higher percentage of patients younger than 50 had BRCA1/2 mutations (22.0% vs. 8.6%).</p>	<p>Large panels yielded a higher VUS rate (28.2% vs. 12.1%) than the guideline panel (10.7% vs. 8.6%). The guideline panel included BRCA1, BRCA2, CHEK2, PALB2, PTEN, STK11, and TP53, ATM, CHEK2.</p>	<p>Patients who do not meet NCCN genetic testing guidelines as reported by their physicians had higher percentages of detectable mutations. Expanded panel testing yields more pathogenic mutations than may be otherwise. Patients older and younger than 50 had different mutation distributions. Expanded panel testing accounts for an important proportion of all pathogenic mutations. Results of this test may be used to inform hereditary mutation study that may be needed. This study supports expanding evidence to the inclusion of candidates who do not meet the requirements of another patient's most common mutation. As test costs continue to decrease, expanded testing will yield a larger group of patients. High confidence panels including BRCA1, BRCA2, CHEK2, PALB2, PTEN, STK11, and TP53, ATM, CHEK2.</p>