Evaluating the NCCN Clinical Criteria for Hereditary Breast and Ovarian Cancer Syndrome

Genetic Testing

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Hereditary Breast and Ovarian Cancer (HBOC) syndrome predisposes females with a \textit{BRCA1} or \textit{BRCA2} mutation to an up to 85% lifetime risk for breast cancer and an up to 40% lifetime risk for ovarian cancer. It is crucial for individuals with HBOC to be identified to allow for proper screening, management, and identification of at-risk family members in order to reduce mortality. The National Comprehensive Cancer Network (NCCN) has established clinical guidelines for when to recommend \textit{BRCA1/2} testing. A retrospective chart review of 1123 M.D. Anderson Cancer Center breast cancer patients was performed in order to evaluate the positive predictive values (PPVs) of 14 individual criterion for predicting a \textit{BRCA1/2} mutation. Two criteria had PPVs significantly below 10%. Only 2 of 115 patients recommended for testing based solely on the criterion of “diagnosed with breast cancer ≤45 years of age” tested positive for a pathogenic mutation at a PPV of 1.6% (0.2-6\%, 95\% CI), which is significantly below the clinical utility cut-off of 10\% (\textit{p} = 0.001). Additionally, 0 out of 37 individuals who underwent testing based on the criterion, “diagnosed with breast cancer at any age with ≥2 close blood relatives with breast cancer at any age” tested positive (0-9\%, 95\% CI). Overall, an individual who meets more than one criterion has a PPV of 12\% while those who meet only one criterion has a PPV of 3.52\%, which is significantly below 10\% (\textit{p}<.0001) for predicting \textit{BRCA1/2} positivity. This data can help provide more personalized risks and anticipatory guidance for patients in their decision to pursue genetic testing.

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