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Expanding *BRCA1/2* testing criteria to include other confirmed breast and ovarian cancer susceptibility genes.

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Background: The National Comprehensive Cancer Network (NCCN) has expanded its breast and ovarian cancer (BC and OC) genetic testing and management recommendations to address the broader spectrum of cancer predisposition genes. However, management recommendations are pending for some candidate BC genes (e.g. *BARD1*, *RAD51D*) and OC genes (e.g. *ATM*, *NBN*) due to insufficient evidence of increased cancer risk, and indications for multi-gene panel testing (MGPT) remain vague overall. We aimed to further characterize BC and OC risks for 21 candidate susceptibility genes and explore the potential utility of *BRCA1/2* testing criteria as an indication for MGPT. **Methods:** Gene-specific pathogenic variant (PV) frequencies among Caucasian BC and OC patients (unadjusted for other personal/family cancer history) ascertained from a cohort of > 175,000 patients referred for MGPT were compared to non-Finnish European reference controls from the genome aggregate database (gnomAD). Clinical histories of BC and OC patients were also reviewed to assess whether NCCN *BRCA1/2* testing criteria were met (version 2.2017). **Results:** We confirmed the association of 15 genes with increased risk (> 2.0-fold) of BC (*ATM*, *BARD1*, *BRCA1/2*, *CDH1*, *CDKN2A*, *CHEK2 (excluding p.I157T)*, *MLH1*, *MSH2*, *MSH6*, *NF1*, *PALB2*, *PTEN*, *RAD51D*, *TP53*). The pooled frequency of PVs in these genes was 9.2% among BC patients of all ethnicities (3.5% in *BRCA1/2* and 5.7% in other genes) and 8.6% among BC patients meeting *BRCA1/2* testing criteria (3.4% in *BRCA1/2* and 5.2% in other genes). Eleven genes were associated with increased risk of OC (> 2.0-fold, *ATM*, *BRCA1/2*, *BRIP1*, *MSH2*, *MSH6*, *NBN*, *PMS2*, *RAD51C*, *RAD51D*, *TP53)*. PVs in these genes were detected among 13.0% of OC patients of all ethnicities (8.0% for *BRCA1/2* and 5.0% for other genes combined). Therefore, inclusion of additional risk genes increases detection rate for BC and OC patients meeting *BRCA1/2* testing criteria by 152.9% and 62.5%, respectively. **Conclusions:** These results further characterize gene-specific BC and OC risks, which can be used to refine management recommendations for at-risk patients. Current testing criteria fail to capture a substantial proportion of women with increased risk of BC and OC.

Title:
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No

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Prevention
Research Category:
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Continued Trial Accrual:
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