Inequitable Access to Genetic Testing Leads to Missed Screening and Prevention Opportunities for Individuals at Risk for Hereditary Breast and Ovarian Cancer

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Background:

The National Comprehensive Cancer Network guideline on Genetic/Familial High Risk: Breast, Ovarian and Pancreatic Cancer (NCCN) recommends offering increased screening and risk-reduction options to women with a pathogenic/likely pathogenic variant (PV/LPV) in 8 clinically-actionable hereditary breast and ovarian cancer (HBOC) genes: *ATM*, *BRCA1*, *BRCA2*, *CDH1*, *CHEK2*, *PALB2*, *PTEN*, and *TP53*. These options range from biannual clinical breast exams and annual breast MRI screening to risk-reducing mastectomy (RRM) and salpingo-oophorectomy (RRSO). Yet health insurance company policies addressing the medical necessity of genetic testing for these genes vary widely. While some payers have adopted the NCCN testing criteria verbatim, others have made specific changes or have developed their own criteria. We sought to quantify the potential impact of this variability on access to enhanced medical management for carriers of PV/LPV in these 8 HBOC genes in a large cohort of patients undergoing genetic testing at a single commercial laboratory.

Methods:

We reviewed clinical and family histories for patients undergoing multigene panel testing that included testing for the 8 genes of interest and identified 107,344 patients who met NCCN testing criteria (excluding prostate cancer criteria, which were not assessed in this study) for HBOC (v.2.2021). We then compared the histories of these patients to testing criteria for 3 different payer groups: Aetna, Blue Shield of California/Federal Blue Cross-Blue Shield, and eviCore (used by over 30 payers including AmeriHealth, Highmark and Horizon). These are the largest payer groups in our cohort and together represent 19% of patients tested. For each patient we determined whether they met testing criteria for the 3 payer groups. For individuals found to have a PV/LPV we determined the potential missed management opportunities for those who would not have been tested under each payer's criteria. In addition, we sought to estimate the maximal impact of this missed management for patients with BRCA PV/LPV (not accounting for patient age or medical history).

Results:

Among patients meeting NCCN testing criteria for HBOC, 10,10,477 (9.8%) were found to have PVs/LPVs in one of the 8 genes. Under the three payer policies 2% to 10.3% of all patients, and up to 4.0% (n=423) of PV/LPV carriers, would not have been eligible for genetic testing. Based on NCCN management guidelines, up to 423 eligible patients would not have been offered annual breast MRI, 208 patients would not have been offered RRM, and 163 eligible patients would not have been offered RRSO.

This lack of testing access due to misaligned medical policies also represents missed opportunities for offering potentially life-saving screening and risk reduction measures to these patients as well. Assuming lifetime cancer risks of 85% for breast cancer and 40% for ovarian cancer, and risk-reduction of 95% with RRM and 80% with RRSO. Had all policies matched NCCN testing criteria, up to 132 breast cancers could

have potentially been detected earlier or prevented and 52 ovarian cancers could potentially have been prevented in our cohort.

Conclusions:

In addition to complicating clinical practice, varying testing guidelines have broader implications. Our data show that a significant number of mutation carriers are being missed by payer policies that deviate from NCCN testing criteria. In turn, this represents missed opportunities to offer proactive screening and risk-reduction options that could potentially save lives for those at risk for hereditary cancer. Based on findings from other studies, these actions would likely reduce health insurer costs as well.