GENETIC COUNSELORS’ APPROACHES TO DIRECT-TO-
CONSUMER GENETIC TESTING FOR HEREDITARY BREAST CANCER

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ABSTRACT

Given the increasing availability of health-related direct-to-consumer genetic testing (DTC-GT) and third-party interpretation (TPI) services, it is likely that genetic counselors (GCs) will continue to encounter consumers that require follow-up counseling for their results. The National Comprehensive Cancer Network recommends clinical-grade genetic testing to confirm commercial results; however, the type of testing that GCs select remains uncharacterized. Therefore, we aimed to describe the specific recommendations that cancer GCs make for confirmatory genetic testing in probands who have already obtained DTC-GT results or TPI data that reported a BRCA1/2 pathogenic variant. We recruited 80 GCs specializing in hereditary cancer and administered a survey that assessed their testing strategy for probands from three hypothetical case scenarios with variable personal and family histories of cancer. The majority of participants would recommend confirmatory clinical-grade genetic testing for both probands’ DTC-GT results and TPI data (77/80, 96%). For probands with a personal diagnosis of breast cancer and a DTC-GT result for an Ashkenazi Jewish BRCA1/2 founder mutation, participants were more likely to recommend targeted testing (single-site or comprehensive BRCA1/2 analysis) (30/77, 39%, p < 0.01). In scenarios where probands had DTC-GT results but lacked a personal and/or family history suggestive of hereditary cancer, and in all scenarios where the probands had positive TPI data for a BRCA1/2 variant, most
GCs would recommend a multi-gene panel. Our results show that GCs are unified in their practice of recommending confirmatory genetic testing, although the selected clinical-grade test varies depending on the proband’s cancer history and type of commercial testing they obtained. As the market for DTC-GT expands and this patient population continues to grow, genetic counselors must continue to be knowledgeable on this topic.