Communication and Information Needs of Women Diagnosed With Ovarian Cancer Regarding Treatment-Focused Genetic Testing

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For women with an ovarian cancer diagnosis, genetic testing for mutations in BRCA1 and BRCA2 has traditionally been limited to those with a significant family history and only after completion of surgery and adjuvant therapy. The greatest benefit of the traditional process, arguably, has been for unaffected family members shown to be at increased risk of breast and ovarian cancer, particularly with regard to breast cancer screening and prevention, and bilateral risk-reducing salpingo-oophorectomy, which has been shown to reduce the risk of ovarian cancer by about 95% (Kauff et al., 2002; Rebbeck et al., 2002). However, new evidence suggests a change in current practice by determining a woman’s mutation status at the time of her ovarian cancer diagnosis and using that information in her treatment plan (Trainer et al., 2010). This genetic testing, offered shortly after diagnosis while a woman’s treatment plan is being considered, is referred to as treatment-focused genetic testing (TFGT).

Support for TFGT stems from preliminary findings that the presence of a germline BRCA mutation defines a genotypic subgroup of epithelial ovarian cancers (EOCs) that have distinct biologic and clinical behavior (Trainer et al., 2010). This behavior has the potential to directly impact treatment and maintenance of ovarian tumors. Importantly, the presence of a BRCA mutation is associated with a better prognosis compared to nonBRCA-related EOCs of similar stage and histologic subtype (Bolton et al., 2012). BRCA-related EOCs are reported to have higher response rates to platinum-based chemotherapy (Chetrit et al., 2008; Tan et al., 2008) and may be less responsive to taxanes than nonhereditary EOCs (Foulkes, 2006; Quinn et al., 2007). In addition, results of phase 1 and phase 2 studies using poly (ADP-ribose) polymerase (PARP) inhibitors (i.e., novel agents that target BRCA-related tumors) in women with other subsets of ovarian cancer have suggested a high risk of ovarian cancer, particularly with regard to breast cancer screening and prevention, and bilateral risk-reducing salpingo-oophorectomy, which has been shown to reduce the risk of ovarian cancer by about 95% (Kauff et al., 2002; Rebbeck et al., 2002). However, new evidence suggests a change in current practice by determining a woman’s mutation status at the time of her ovarian cancer diagnosis and using that information in her treatment plan (Trainer et al., 2010). This genetic testing, offered shortly after diagnosis while a woman’s treatment plan is being considered, is referred to as treatment-focused genetic testing (TFGT).

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