Getting to the Point: What Women Newly Diagnosed With Breast Cancer Want to Know About Treatment-Focused Genetic Testing

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Traditionally, genetic testing for a germ-line mutation in the breast cancer protection genes (i.e., BRCA1 or BRCA2) in women with a breast cancer diagnosis has been confined to those with a family history of breast and/or ovarian cancer. Testing of such women usually takes place after completion of active cancer treatment with the purpose of guiding their future risk management in relation to a new primary breast and/or ovarian cancer diagnosis. In addition, genetic risk information has significant risk management implications for a woman’s unaffected genetic relatives who may themselves consider predictive genetic testing to clarify their risk of breast and/or ovarian cancer.

However, growing evidence shows that mutation status may influence breast cancer management recommendations and that benefits may arise from having genetic counseling and testing available shortly after a cancer diagnosis (Silva, 2008; Trainer et al., 2010; Tutt & Ashworth, 2008). The process, referred to in this article as treatment-focused genetic testing (TFGT), may help guide a woman’s initial cancer treatment and future risk management. In the first instance, TFGT may assist with the complex decision-making process regarding a woman’s surgical options, including the selection of breast conservation or a therapeutic mastectomy, with or without a contralateral risk-reducing mastectomy, based on the patient’s mutation status and/or family history of cancer (Meiser et al., 2008).

In addition, evidence suggests that BRCA-associated tumors are more sensitive to platinum agents and potentially less resistant to taxanes (Quinn et al., 2007; Rottenberg et al., 2007; Trainer et al., 2010). Novel agents also are being tested that target BRCA tumors;