

Genetic Testing After Previous *BRCA* Testing: A Case Study

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Mutations linked to hereditary cancer syndromes may increase an individual's risk of developing cancer, as well as its recurrence. New genes that may also carry pathogenic mutations associated with cancer risk have been identified; as a result, individuals previously tested should consider additional testing. This article provides a case study illustrating the importance of such testing.

At a Glance

- Although just a small percentage of cancers have a genetic link, individuals identified as having pathogenic mutations may develop cancers earlier—and cancers that are more aggressive—than the general population.
- Oncology nurses are often among the first to discuss with patients their fears regarding the risks of cancer development in future generations.
- Identifying a pathogenic mutation early can assist in cancer prevention or earlier detection.

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About 5%–10% of all cancers are related to a genetic mutation inherited from an individual's parents (National Cancer Institute, 2013). These identified mutations are related to hereditary cancer syndromes that may significantly increase an individual's risk of primary and secondary cancers, as well as cancer recurrences (Yurgelun et al., 2015). In 2013, the U.S. Supreme Court ruled against the ability to patent specific genes (*Association for Molecular Pathology v. Myriad Genetics, Inc.*, 2013); as a result, genetic testing for cancer risk exploded because more than just one third-party lab could perform *BRCA1/2* testing. The growing availability of genetic testing through reduced costs and improved insurance coverage, as

well as the continued identification of new genes related to cancer risk, raises the possibility of multiple pathogenic variants in a single individual. Individuals previously tested for *BRCA1/2* mutations may now need to consider additional testing because of the identification of new genes that may also carry pathogenic mutations associated with an increased risk of cancer.

The National Comprehensive Cancer Network ([NCCN], 2016) provides guidelines and recommendations for testing individuals for hereditary breast and ovarian cancer (HBOC). The guidelines in Figure 1 depict very specific criteria for practitioners to identify when considering genetic testing. Although recommendations are available for commonly known genes or those that

pose a significant increase in cancer risk, recommendations are still lacking for testing individuals with extensive cancer histories on both sides of the family or even within the same side.

Genetic counselors are in demand and hard to find throughout the United States, except in larger cities or commercial laboratories. Therefore, training primary care providers (physicians, nurse practitioners, clinical nurse specialists, physician assistants, and nurse midwives) who plan to order genetic testing to understand that no case or family history is the same and can often be more involved than what is expected is imperative. For instance, with the introduction of multigene panel testing, an individual with a strong family history of breast cancer may exhibit a genetic mutation in an unexpected gene, such as *CDH1*, which puts the individual at an extremely high lifetime risk of gastric cancer, in addition to the breast cancer risk. The following case study will illustrate how S.L., a patient known to be positive for HBOC, was previously managed based solely on her known *BRCA2* mutation. However, after discussion with and evaluation by a genetic practitioner, she was determined to be qualified for additional testing; ruling out any other potential pathogenic mutations was also thought to be prudent.

Case Study

S.L. is a 41-year-old female patient presenting to the clinic to discuss her known pathogenic mutation in *BRCA2*. She was diagnosed with invasive lobular carcinoma of the left breast at age 33 years; afterward, she underwent bilateral total mastectomy with reconstruction