

Tumor genomic testing is used primarily to facilitate the selection of the best possible treatment for a malignancy based on the genomic characteristics of the tumor. Germline genomic testing has implications for care and recommendations for cancer prevention and early detection for the patient and their family. Careful review of specific components of tumor genomic testing reports and of the family history of malignancy can help ensure that families with potential germline risk are identified and referred for genetic counseling and genetic testing.

AT A GLANCE

- Tumor genomic testing can provide information about what might be the best treatment for a malignancy and the potential germline risk for developing a malignancy.
- Clinical nurse competency includes the ability to understand a tumor genomic testing report, which lists possible indicators of germline risk.
- The identification of patients and families with potential germline risk, as well as referral of these families for genetic counseling and testing, can facilitate implementation of the best possible recommendations for cancer prevention and early detection for patients and family members.

KEYWORDS

tumor genomic testing;
germline genomic testing;
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DIGITAL OBJECT

IDENTIFIER

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Tumor Genomic Testing

Identifying characteristics associated with germline risk for developing malignancy

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Germline testing to determine if an individual and other family members have a hereditary risk for developing a malignancy has been used in oncology care for two decades. Germline pathogenic variants (historically referred to as mutations) occur in the egg or sperm and can be passed from parent to offspring. In the 2000s, germline testing for pathogenic variants in the *BRCA1/2* genes was available; additional genes have been gradually identified. Germline testing typically includes a panel of 40 or more genes known to be associated with an increased risk for developing cancer. Germline pathogenic variants are thought to be related to the development of malignancy in 5% to 10% of all diagnosed cases (National Comprehensive Cancer Network [NCCN], 2020a). Germline testing provides information that can lead to cancer prevention, including risk-reducing surgery and the early detection of a malignancy when treatment can be most effective.

Genomic testing for pathogenic variants and biomarkers in a tumor can lead to the selection of an optimal treatment (Ngeow & Eng, 2016). Tumor genomic testing focuses on many somatic, or acquired, variants in the tumor. These are not passed from generation to generation. For example, for more than a decade, the identification of non-small cell lung tumors that overexpress epidermal growth factor receptor (EGFR) has enabled the selection of an EGFR inhibitor therapy that is more likely to be an effective treatment (Ngeow & Eng,

2016). Many of these acquired, or somatic, pathogenic variants are not known to be associated with germline risk and do not have implications for other family members.

Intersection of Tumor and Germline Genomic Testing

Tumor genomic testing and germline genomic testing are part of personalized medicine, which aims to select the best recommendations for care based on the genomic makeup of the individual and/or tumor. Traditionally, these have been considered to be two discrete types of genomic testing; however, advances in genomic science have demonstrated that there is overlap between them. There are no guidelines for the identification of germline risk based on tumor genomic testing (Clark et al., 2019). According to the American Society of Clinical Oncology (ASCO), oncology providers should inform patients that there is the possibility of incidental and secondary germline findings when tumor testing is performed so that the patient and family members are prepared for results that are suggestive of potential germline risk (Robson et al., 2015).

Tumors may have germline and/or acquired, or somatic, pathogenic variants. Both have implications for the selection of the best possible treatment (Aldredge & Randall, 2019). In some cancer institutions in the United States, healthcare providers have access to germline and tumor genomic testing with the goals of selecting the best possible