

Using direct-to-consumer genetic testing (DTCGT), individuals can order a genetic test, collect and submit a saliva sample, and obtain results about their genetic risk for a variety of traits and health conditions without involving a healthcare provider. Potential benefits of DTCGT include personal control over genetic information and health management decisions, whereas potential risks include misinterpretation of results, psychosocial distress, and lack of informed consent. Oncology nurses can provide education, support, and advocacy to enable patients to truly understand the positives and negatives associated with DTCGT.

#### AT A GLANCE

- DTCGT is readily available and can provide limited information about risks for developing various common diseases and traits, as well as ancestry information.
- Such testing is typically completed without counseling and guidance from a knowledgeable genetics professional.
- DTCGT often does not involve comprehensive sequencing of multiple genes associated with risk for developing malignancy, and it accounts for a small percentage of genetic changes associated with an increased risk for developing malignancy or other diseases.

#### KEYWORDS

direct-to-consumer genetic testing; genetic counseling; psychosocial distress

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# Direct-to-Consumer Genetic Testing

Helping patients make informed choices

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Oncology nurses are likely to encounter patients who have questions about direct-to-consumer genetic testing (DTCGT), which is genetic testing sold directly to consumers (often via the Internet) that can provide ancestry information and predict risk for multiple common diseases and traits. DTCGT usually occurs without the involvement of a knowledgeable healthcare professional. Advertisements and quality regulatory oversight for DTCGT are limited, but it accounts for an estimated \$230 million in business each year (Brierley, Bonadies, Moyer, & Matloff, 2014; Su, 2013). Oncology nurses need to be aware of the strengths and limitations of DTCGT to provide accurate patient education and support.

## Types of Direct-to-Consumer Genetic Testing

DTCGT differs from traditional genetic testing because it is available outside of the context of a healthcare professional's evaluation and is accessible to all, regardless of whether they satisfy the traditional criteria for genetic testing (Gollust et al., 2017). Individuals who undergo DTCGT submit a saliva sample obtained in their home and receive genetic risk results (typically via an email) for multiple conditions, including malignancy, heart disease, and dementia. Pricing ranges from about \$100–\$1,000, depending on the extent and type of testing (Su, 2013).

DTCGT uses single-nucleotide polymorphisms (SNPs), which are genetic

variations that represent a difference in a single nucleotide in the DNA. Unfortunately, the SNPs employed in DTCGT account for just a small percentage of inherited risk factors, and the risk predictions based on SNP profiles often have unclear clinical implications (Wynn & Chung, 2017). SNPs look only at pieces of DNA, which is much different than sequencing an entire gene by examining the full sequence of nucleotides.

DTCGT includes genetic ancestry testing known as genetic genealogy. DNA variations can provide information about where an individual's ancestors may have originated and about relationships between families. The Y chromosome is passed exclusively from father to son and is tested to explore ancestry in the male line, whereas mitochondrial DNA testing, which is passed from the mother, is used to provide information about the maternal line. The SNPs of an individual who undergoes testing are also compared with the SNPs of those previously tested to link relatives together (Gray et al., 2017).

## Governmental Regulatory Oversight

Regulatory oversight of DTCGT is currently limited, fragmented, and not under the auspices of any one agency (Agurs-Collins et al., 2015). The U.S. Food and Drug Administration has the authority to regulate the safety, efficacy, and security of human drugs, biologic products, and medical devices, and the Federal Trade Commission has the authority to regulate advertising of health-related information