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.

**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...
to consumers, ensuring that it is not false or misleading. Under the Clinical Laboratory Improvement Amendments of 1988, the Centers for Medicare and Medicaid Services regulate clinical laboratories but do not examine whether the tests performed there are clinically meaningful.

**Potential Benefits and Risks**

Proponents of DTCGT acknowledge that it allows individuals to pursue testing independently and may increase personal responsibility for health and lifestyle choices (Gollust et al., 2017). The individual undergoing such testing also has a measure of control over his or her privacy and what information is included in medical records. DTCGT also raises public awareness of the role of genetics in health and healthcare. However, despite the benefits, DTCGT contains a number of potential risks.

**Informed Consent**

A major component of formal genetic counseling includes providing adequate information so that informed consent is possible. Individuals need to understand the strengths, limitations, and risks associated with genetic counseling (Mahon, 2013). In DTCGT, information about the test may be difficult to discern because the risks and limitations of testing are often described in the fine print and not footnotes on the website. A content analysis of 23 health-related DTCGT websites demonstrated that information about the benefits of the testing outweighed information about its risks and limitations by a ratio of 6 to 1 (Singleton, Erby, Foisie, & Kaphingst, 2012). Commonly cited benefits include prevention of disease, personalized medical recommendations, and personal control over health. Risks cited in the 23 studies reviewed include anxiety or worry (6 studies), genetic discrimination (4 studies), and negative impact on family members (1 study). Two studies cited no risks. What is concerning is that many individuals using DTCGT do not realize that it is not comprehensive genetic testing and that negative test results do not mean that an increased risk for developing malignancy or other disease does not exist (Gollust et al., 2017). Other risks that are not usually explicitly described include misinterpretation of results or distortion of the clinical significance of the results to overall health. Misinterpretation of the significance of the results on the part of the user may lead to failure to engage in preventive behaviors because the risk is not adequately understood or is underestimated. Conversely, the individual who has undergone testing may engage in inappropriate and potentially harmful health-related action if the risk is overestimated.

In a study comparing the genetic test ordering habits of nongenetics professionals with cancer genetics professionals, the latter were significantly less likely to order their own testing, which suggests that even those who presumably know the most about counseling and testing regarding cancer genetics realize they would need guidance and support from an unbiased, knowledgeable professional (Brierley et al., 2014).

**Case Study 1**

A family with a known CHEK2 mutation sought testing. The insurance held by the family member to be tested would not cover the testing, and the family believed that the price quoted for out-of-pocket testing was too high. They pursued DTCGT, which included a cancer panel, and the family called and stated they were relieved the family member was negative. Unfortunately, the CHEK2 mutation that the family harbored was not on the panel. The test panel included only SNPs for some BRCA1/2 mutations. The result was noninformative, but the family believed they had received the answer they were looking for because the panel included “cancer genes.” They failed to understand that it was not targeted testing or comprehensive sequencing of genes associated with an increased risk for developing cancer.

**Psychosocial Distress**

Psychosocial distress for the individual and the family following DTCGT is another concern. The combination of high perceived seriousness for developing a disease and a DTCGT result suggesting increased risk for a serious disease, such as cancer, heart disease, or dementia, can be associated with an increase in distress (Barton, 2017). In addition, misattribution of paternity is regularly discussed prior to genetic testing with a genetics professional. However, with DTCGT, this does not occur, often resulting in the significant emotional distress that accompanies an individual learning that he or she has a different parent (Stoeklé, Mamzer-Bruneel, Vogt, & Hervé, 2016).

**Case Study 2**

A couple decides to undergo DTCGT primarily for entertainment purposes, and the woman learns she has one of the three founder mutations in the BRCA1/2 gene, which are common in those of Ashkenazi Jewish ancestry. The woman, who previously thought her risk for developing

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"Regulatory oversight of direct-to-consumer genetic testing is limited, fragmented, and not under the auspices of any one agency."
breast cancer was similar to that of the
general population, has suddenly learned
that her breast cancer risk may be as high
as 85% and that her ovarian cancer risk
may approach 50%. No anticipatory guid-
ance was offered for this possible result,
which can be stressful and accompanied by
many difficult decisions regarding
risk-reducing surgeries.

Specimen Retention
Although DTCGT also often includes
a research component in that the data are
used for further research and the
development of additional panels, this in-
formation is frequently buried in the fine
print of the consent to test, alongside pay-
ment information. Consequently, patients
may or may not realize they are consent-
ing to allow their specimen to be used
for further research (Stoeklé et al., 2016).
When genetic counselors obtain informed
consent prior to testing, they have a pa-
tient consciously check or not check a box
to identify whether they want their speci-
men used for further research.

Implications for Nursing
and Conclusion
Singleton et al. (2012) reported in their
case analysis that 70% of the 23 DTCGT
websites examined recommend that
consumers discuss the findings with a
healthcare provider. However, a primary
care provider may not have enough ge-
netic expertise to interpret the results
and may not realize the inability of a few
SNPs to accurately predict disease risk
(van der Wouden et al., 2016). Oncology
nurses need to be knowledgeable about
the strengths and limitations of DTCGT
and acknowledge that genetic assess-
ment can be beneficial in making good
healthcare decisions. By paying attention
and being receptive to a discussion on
 genetic risk and DTCGT, nurses have the
opportunity to educate patients about
DTCGT and genetic risk. Patients should
feel comfortable sharing DTCGT results
with healthcare providers. When appro-
 priate, nurses need to refer patients to a
 genetics professional for more in-depth
counseling, testing, and interpretation of
results. Patients who do not believe that
healthcare professionals are adequately
addressing their genetic risk by inquiring
about family history, addressing concerns
about genetic risk, or referring the patient
to a genetics professional may attempt
to gain a better understanding through
DTCGT, which may be inadequate (Burke
& Trinidad, 2016). Many resources exist
for oncology nurses and patients to learn
more about genetic testing and DTCGT
(see Figure 1).

Nurses need to actively inquire about
whether patients have concerns about
hereditary risk, and they must know how
to refer patients and their families to

credentialed genetics professionals for
comprehensive evaluation, counseling, se-
lection of the best tests to evaluate genetic
risk, and test interpretation, including
management recommendations. In addi-
tion, nurses can educate patients on the
benefits of comprehensive genetic care
and the potential limitations of DTCGT.

A long-standing tenet of genetic testing
is that, in the pretest phase, it is important
to understand what an individual intends
to do with the results and to communicate
that the results of such testing can poten-
tially affect the individual’s entire family.
DTCGT may not be able to provide the
information a patient or family is seek-
ing (Carere, Kraft, Kaphingst, Roberts,
& Green, 2016). For instance, a patient may

FIGURE 1.
DIRECT-TO-CONSUMER GENETIC TESTING (DTCGT) RESOURCES

AMERICAN COLLEGE OF MEDICAL GENETICS
Policy statement that recommends that a genetics professional be involved in the testing process
and that the consumer be fully informed of the
strengths and limitations of DTCGT; privacy issues
should also be addressed.

AMERICAN SOCIETY OF HUMAN GENETICS
Policy statement that recommends transparency
in DTCGT; calls for education of healthcare
professionals on DTCGT and oversight of the
validity of the test, laboratory testing, and
reporting methodology; and states that genetic
testing should include counseling by a genetics
professional

ASSOCIATION FOR MOLECULAR PATHOLOGY
Position statement that recommends business
transparency and that testing be clinically
meaningful and consumers be informed of the
limitations of DTCGT

CENTERS FOR DISEASE CONTROL AND PREVENTION
Consumer discussion of the strengths and
limitations of DTCGT

FEDERAL TRADE COMMISSION
Consumer guidance on the strengths and
limitations of DTCGT

GENETIC ALLIANCE
- Consumer information on the regulation of DTCGT

INTERNATIONAL SOCIETY OF NURSES IN GENETICS
Position statement that recommends balanced
discussion of risks and benefits, as well as
implications for nursing, in clinical practice

NATIONAL SOCIETY OF GENETIC COUNSELORS
Consumer-oriented website on facts about
DTCGT, as well as its strengths and limitations

SMITHSONIAN
Interactive website that describes ancestry genetic
testing

AMERICAN STATEMENT THAT RECOMMENDS BUSINESS
TRANSPARENCY AND THAT TESTING BE CLINICALLY
MEANINGFUL AND CONSUMERS BE INFORMED OF THE
LIMITATIONS OF DTCGT

Interactive website that describes ancestry genetic
testing
undergo testing for some BRCA SNPs and, based on these results, think that she does not have an elevated risk for developing breast or ovarian cancer. The patient may then underestimate the significance of her family history, thinking she had comprehensive testing for genetic risk; in reality, however, her testing was very limited and involved only a few genes associated with an increased risk for developing breast and/or ovarian cancer. The issue stems from a lack of understanding concerning the differences in comprehensive multi-gene sequencing versus SNP-based testing (Barton, 2017).

Patients often state that they engage in DTCGT because they want to make better decisions regarding their lifestyle habits (Barton, 2017). Educating patients about the benefits of a healthy lifestyle is important, regardless of genetic testing results. When patients feel that healthcare providers are not paying attention to genetic risk or wellness issues, they are more likely to turn to DTCGT, thinking it will provide answers (Burke & Trinidad, 2016). Oncology nurses need to take the time to discuss with patients their perceptions and concerns related to genetic risk and provide encouragement, support, and education on the importance and role of a healthy lifestyle in reducing cancer risk. In addition, oncology nurses can make referrals to genetics professionals for comprehensive risk assessment and appropriate testing based on risk. They can also provide education and support to help patients make informed decisions about the strengths, risks, and limitations of DTCGT.

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The author takes full responsibility for this content and did not receive honoraria or disclose any relevant financial relationships.

REFERENCES


DO YOU HAVE AN INTERESTING TOPIC TO SHARE?

Genetics & Genomics aims to educate oncology nurses about the emerging role of genetics and genomics in cancer care. If you are interested in writing for this department, contact Associate Editor Suzanne M. Mahon, DNSc, RN, AOCN®, AGN-BC, at suzanne.mahon@health.slu.edu.