Position Statement: Direct-to-Consumer Genetic/Genomic Testing (DTC-GT)

Background

Direct-to-consumer genetic/genomic testing (DTC-GT) is defined as marketing and/or provision of genetic/genomic tests directly to the public, generally without the involvement of healthcare providers (Genome.gov, 2022). Currently, DTC-GT for personalized information about health status, disease risk, and other traits includes testing that detects variants in one or several genes (e.g., for a specific condition), targeted genome panels (e.g., cancer syndromes), and whole genome scans or sequencing (e.g., for broad-based risk assessments based on many genetic variants). Demand for DTC-GT as well as rising public awareness across the globe are expected to expand (Global Marketing Insights, 2022). According to the National Human Genome Research Institute:

> The global "direct-to-consumer" (or DTC) genomic testing market was valued at $117 million in 2017, suggesting millions of people are purchasing these tests. In contrast to clinical genomic testing (such as for cancer [see **Cancer Genomics**] and prenatal screening [see **NIPT**]), in which a healthcare provider orders the tests and provides the results, DTC genomic tests can be purchased online or even at your local pharmacy (Genome.gov, 2022)

This leaves much to the direct consumers, who purchase such testing kits, up to their own interpretation and belief systems. There is even an implicit requirement for consumers to continue monitoring the site for updates because evolving knowledge from DTC-GT data bases change as more genetic profiles are added. On one hand, direct-to-consumer genetic/genomic testing empowers individuals to pursue genetic/genomic testing and information independently, make autonomous decisions, and take personal responsibility for health, healthcare, and lifestyle choices (Wessel et al., 2016). It also provides consumers the privacy and control of personal genetic health information, including its entry into individual medical records (van der Wouden et al., 2016). On the other hand, the DTC-GT environment shifts the driving force behind access to personal genetic information from the healthcare professional to the consumer (American College of Medical Genetics and Genomics [ACMG], 2016; American College of Obstetricians and Gynecologists [ACOG], 2021). Clinicians who may have little to no background in genetics and genomics are expected to opine knowledgeably about DTC-GT results in episodic conversations with patients.

Consensus on the impact of DTC-GT is still evolving and therefore definitive evaluations would be premature. In this context, providers of healthcare services should be up to date with potential benefits, risks and limitations of DTC-GT in order to promote informed professional and individual health-related decisions. This position statement is intended to provide nurses, as well as other healthcare providers, policymakers, and the general public, with information and key

factors relevant to informed decision-making about the use of DTC-GT. The focus of this position statement is on DTC health-related genomic testing such as disease risk assessment, drug response genetic typing, and carrier testing for recessive conditions that are initiated without the order of a healthcare provider. As such, this statement does not address reasons outside the parameters of health (e.g., ancestry, trait characteristics, paternity testing).

Issues

Informed consent. Genome testing and the interpretation of genetic test results are highly complex, technical and limited by the extent of scientific knowledge about individual variants. The consumer should be fully informed regarding the purpose, extent, and intent of genome testing, its scientific validity and clinical utility, and what the tests can and cannot determine about their health (ACMG, 2016). In DTC-GT, the degree to which consent is informed is limited by consumers' general and health literacy and numeracy skills. Online resources for consumers tend to lack essential information for proper informed consent (Niemiec & Howard, 2016) without provider's anticipatory guidance. While significant improvements have occurred in the way results are communicated to enhance consumer understanding, significant gaps remain in consumers' ability to interpret findings (Ostergren et al, 2015).

Informed consent for children, especially newborns, requires particular attention because young children cannot assent or consent for themselves. A complicated patchwork of law across the U.S. from state-to-state and national standards around the world define the legal landscape regarding consent of minors. However, vigorous marketing to parents is creating mounting pressure to have their offspring screened by either whole genome sequencing (WGS) or whole exome sequencing (WES). These forms of testing provide abundant, and sometimes incorrect, misinterpreted, or misleading information regarding children's health. Nurses in all settings that provide services to children or pregnant persons should be prepared to answer questions about WGS or WES for children or make appropriate referrals. The American Academy of Pediatrics strongly discourages DTC-GT for children (Ross et al, 2013).

Validity of findings. Regulation of clinical laboratory testing, and in particular DTC-GT, varies according to national policy and laboratory standards (Rafiq et al. 2015), as do consumer protections that oversee marketing to the public (genome.gov, n.d.). In clinical genomics laboratories, safeguards include laboratory quality standards, provider involvement, restrictions on tests offered, clinical standards, informed consent processes, and marketing regulations. DTC-GT companies differ in their approaches to these and other factors that assure validity of results, including health care provider involvement, genetic counseling services, testing methodologies and foci, and provision of information; these factors and safeguards should be assessed before proceeding with DTC-GT (genome.gov, n.d.). Furthermore, historical cases of DTC-GT service providers reporting false positives (Tandy-Connor et al, 2018) and other incorrect information have highlighted the potential for error in determining or reporting results to consumers (Nelson et al, 2019). Therefore, consensus states that clinically meaningful results should be considered unreliable until confirmatory clinical genetic testing is obtained (genome.gov, n.d.).

Misinterpretation of test results. Misinterpretation of results or distortion of the extent of significance to the overall health or risk status of the person tested may occur when genomic testing is performed without consultation from healthcare providers. Individuals who use DTC-GT have varying levels of health and general literacy and numeracy, with many individuals unable to accurately interpret findings or know when to seek follow-up healthcare (Tolan, 2020). Misinterpretation may lead to inappropriate and potentially harmful health-related action or inaction. Furthermore, results may identify variants of uncertain significance, thus posing challenges to interpretation (Jackson et al., 2014). Additionally, there are challenges in application to healthcare based on the personal and family histories that prompted the testing.

Psychosocial concerns: The potential for psychological harm has also been raised (Covolo et al., 2015). Although research in adults suggests some negative psychological impacts (e.g., anxiety) in some DTC-GT users, these effects appear to be short-lived (no significant differences by one year after testing), and personal value has been attributed to testing, regardless of results (Covolo et al., 2015). However, social, familial, and anticipatory guidance considerations may affect individuals related to matters of paternity, adoption, and so on, with potentially fraught psychosocial effects (Moray et al, 2017).

Confidentiality, privacy & integrity of specimens: Confidentiality and integrity of genomic information are also concerns in DTC-GT as they are in every aspect of healthcare in order to maintain ethical practice. Many companies rely on free access to individual testing data to populate their internal databases for comparison with others and may share this information without specific consent or releases from customers (Genome.gov, 2022). Sharing and resale of customer data has included personally identifying information and data delivery with other commercial entities as well as governmental actors including law enforcement agents (Hendricks-Sturrup et al, 2019). Documentation, policies, regulations and security measures for the protection and privacy of consumers and their test samples, for assuring the quality and reliability of testing, for the handling, storage and reuse of samples, and for marketing, is imperative (ACMG, 2016). Appropriate oversight is being addressed, yet with recognized gaps (Food & Drug Administration [FDA], 2015; FDA, 2017).

Recommendations

Therefore, it is the position of the International Society of Nurses in Genetics that nurses should:

1. Be informed about DTC-GT and associated health, ethical, legal, and social issues, together with updated information of genetic evaluation, technology, and services related to one's professional scope of practice.

2. Be receptive to communication with patients about DTC-GT to explore their expectations regarding the personal value of genomic information and its intended use.

3. Foster consumer understanding by educating about: (a) the role of genes and environment in health and disease, (b) the importance of family history, (c) benefits and limitations of DTC-GT, and (d) genetic risk assessment, risk reduction, disease prevention, and health promotion options.

4. Facilitate access to appropriate genetic services for those with unaddressed genetic risks or concerns, and for those planning to make changes in health behavior or family planning based upon information provided by DTC-GT.

5. Evaluate patients' results in greater depth based on clinical genetic testing labs and obtain detailed medical and family histories with the patient and other family informants when appropriate.

6. Clinically confirm DTC-GT results that lead to clinical decision-making in a certified genetics laboratory when possible.

7. Consult with and if indicated, refer patients to clinical geneticists and pharmacists knowledgeable about pharmacogenomics and other health-care professionals (such as nutrition and developmental specialists) when indicated (NHGRI, 2022).

8. In addition to the above recommendations, nurse educators are advised to provide a foundation in genetics/genomics at the undergraduate level and to foster more advanced genetics/genomics knowledge, understanding, assessment skills, and awareness of ethical, legal, professional, regulatory and policy issues at the graduate level. Nurses should be able to integrate these considerations into health care practice, including persons interested in or using DTC-GT. Nurse researchers are encouraged to investigate the physical, psychological, social, and economic effects of DTC-GT and to identify strategies that foster optimal delivery of genetic information and services.

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