Executive Summary

Direct-to-consumer (DTC) genetic tests may be perceived by the public as providing both clinically valuable information, as obtained from medical devices; and general educational value, as obtained from devices for recreational purposes. DTC genetic tests, when regarded as medical devices, are problematic for several reasons (personal communication; Roundtable, July 17, 2017). First, these genetic tests are often carried out in laboratories that are not held to industry-level standards for clinical genetic testing. Second, DTC genetic testing can exploit consumers by neglecting to disclose the true degree of the clinical utility of the test and the meaning of the test results within the context of individual and familial risk factors. Third, DTC genetic testing can result in a higher rate of false positives, anxiety over results, as well as risks and additional costs of downstream retesting or interventions based on false-positive testing. Lastly, privacy of individual genetic data is a concern, as for-profit testing companies may sell information, potentially resulting in discriminatory practices by insurers and others. The American Academy of Nursing (Academy) supports efforts to ensure that genomic technologies are applied through appropriate Federal Drug Administration (FDA) and Federal Trade Commission (FTC) oversight, statutory regulation of genetic testing, and legislation to prevent genetic discrimination. The Academy will work with our affiliates and consumer organizations to increase nurses’ role in genetic education for patients and families and to disseminate best practices for genetic testing for health-care providers and consumers.

Background

Genetic testing has traditionally been ordered by health-care providers to identify risk, diagnose, or treat an illness; however, with the advent of new technologies that greatly decrease test costs, genetic tests are increasingly being marketed to individual consumers. This approach presents a myriad of problems related to clinical applicability, validity, and reliability of testing, as well as potential for genetic discrimination if data are not protected. DTC testing is of concern for clinicians, consumer groups, and policy makers, yet no firm legislative policy has been enacted to ensure that individuals are provided with transparent information on the limitations of genetic testing for clinical purposes, appropriate interpretation of results, potential loss of privacy, and alternative options for testing (Kaufman, Bollinger, Dvoskin, & Scott, 2012).

Genetic testing includes an array of techniques for analyzing human DNA, RNA, and protein. These services range from detecting gene variants associated with a specific disease, such as Alzheimer’s and Huntington’s disease, to paternity testing. Genetic tests are currently under regulatory control by the U.S. Department of Health and Human Services, Centers for Medicaid and Medicare Services (CMS), FDA, and FTC. CMS is responsible for regulating all clinical laboratories performing genetic testing, ensuring the laboratory’s compliance with the Clinical Laboratory Improvement Amendments of 1988 (CLIA). The FDA’s role is focused on regulating the safety and effectiveness of genetic tests as medical devices, and their role depends on how the test comes to market. The test may be marketed as a commercial test “kit” or as a laboratory-developed test (LDT). A
commercial test kit is a group of reagents that are packaged together and sold to other laboratories, and in this case, the FDA has maintained consistent oversight on the safety and effectiveness of commercial kits. In contrast, an LDT is developed and performed by a single laboratory; all specimens are sent to the laboratory, and the company maintains a monopoly over the interpretation of the test and may or may not disclose what will happen to the sample or the sample results to the consumer. As a general rule, the FDA has practiced “enforcement discretion” for LDTs based on the category of the test. Category 3 tests generally receive some oversight by the FDA, and include “high-risk” genetic tests that provide information with direct implications on the individual’s risk of mortality. Other categories of tests have generally received little attention from the FDA. The FTC’s regulatory authority is focused on how genetic tests are advertised with the primary goal of preventing false advertisement or misleading statements.

Many DTC tests are carried out in laboratories that are not adherent to CLIA standards. The purpose of CLIA is to certify the clinical testing quality, including verification of the procedures used and the qualifications of the technicians processing the tests. There is little assurance of validity (accuracy of test) or reliability (repeatability of results) for tests carried out in non-CLIA laboratories. In addition, DTC testing companies do not address whether minors are able to participate in testing, and most policies do not mention the vulnerability of receiving unexpected information by individuals and family members (Moray, Pink, Borry, & Larmuseau, 2017). Furthermore, most companies do not meet international transparency guidelines related to confidentiality, privacy, and secondary use of data (Laestadius, Rich, & Auer, 2017). DTC tests seldom are based on published guidelines for appropriateness of testing to guide clinical decisions regarding risk of, diagnosis of, or treatment of disease (Rockwell, 2017).

Although the FDA has been actively providing guidance to some commercial LDT and DTC companies, others have proliferated without any scrutiny. With currently 84 competitors and a projected world market for genetic testing forecast to reach $7.4 billion by 2020, there is a high risk of decisions regarding oversight to be influenced by economics rather than evidence on analytic and clinical validity (Global Industry Analysts, Inc., 2017). Further, the costs in terms of anxiety and follow-up clinical tests and procedures are unknown, and follow-up tests are not covered by DTC companies. As the FDA does not evaluate clinical utility, there are already tests on the market that have been criticized for increasing costs but not improving care or patient outcomes (Wallace, 2017). In addition, recent data evaluating the influence of DTC testing on individual health decision making have shown little impact in improving health promotion and disease prevention activities of consumers (Barton, 2017; Nielsen, Carere, Wang, Roberts, Green, PGen Study Group, 2017).

Further, consumer expectations may not be realized. Patients are given results without the benefit of consultation with a health-care professional who can interpret the results and make appropriate clinical recommendations based on the individual’s current health status, lifestyle, and family health history. Low-risk patient populations, who are more likely to engage in DTC testing and are statistically at increased risk to receive false-positive results, are likely to provoke a cascade of additional downstream interventions, generating additional costs, when they inform their healthcare provider about positive testing results (Kaufman et al., 2012). Public or private insurers, or the patient, are then left paying for any office visits or downstream interventions even though the health-care provider would not have initiated the testing cascade. Thus, the private DTC companies avoid accountability for the unnecessary medical testing they provide, but realize the profits. In addition, consumers may be placing themselves at risk of genetic discrimination in the future. Proposed federal legislation, such as H.R. 1313, highlights the potential danger for individuals and families to experience genetic discrimination by employers and health insurance companies (Hudson & Pollitz, 2017; Pollitz & Rae, 2017).

### Response and Policy Options

As early as 1997, government advisory panels have been calling for FDA oversight of LDTs and review of the analytic and clinical validity of genetic tests before allowing them to go to market (National Institutes of Health-Department of Energy Working Group on Ethical, Legal and Social Implications of Human Genome Research, Task Force on Genetic Testing, 1997; Secretary’s Advisory Committee on Genetic Testing, 2000). A 2006 investigation by the Government Accountability Office reported that many of the genetic DTC tests were medically unproven, meaningless, and misleading for consumers. Another report by the Secretary’s Advisory Committee on Genetics, Health and Society in 2008 recommended that the FDA regulate all genetic testing (Ferreira-Gonzalez et al., 2008). In 2010, the Government Accountability Office issued another report concerning the deceptive practices of DTC genetic testing, which were summarized as “misleading and of little or no practical use” (U.S. Government Accountability Office, 2010, p. 4).

Although federal legislation has been introduced that would increase FDA oversight to all clinical laboratory testing and the regulation of genetic tests, the legislation lacked necessary support to be enacted into law. The Laboratory Test Improvement Act was coauthored in 2006 by Senator Edward Kennedy (D-MA) and Senator Gordon Smith (R-OR) to expand FDA oversight of genetic tests. In the same year, then-Senator Barack Obama (D-IL), with cosponsorship from Senator Richard Burr (R-NC), introduced the Genomics and Personalized Medicine Act, with provisions to strengthen the regulation of genetic testing (S. 3822). The bill was introduced again
in 2007 and revised and reintroduced in 2010 (S.976, S. 5440). A major reason that the legislation was not passed was fear that too much oversight might impair medical progress and, perhaps, curtail commercial growth (Rockwell, 2017).

Another avenue for addressing this issue has been through state legislation that restricts laboratory testing to the orders provided by a licensed health-care professional. Currently, only a small number of states—including Connecticut, Georgia, Hawaii, Idaho, Kentucky, New Hampshire, Pennsylvania, Rhode Island, and Tennessee—explicitly require the order of a physician or other licensed health-care professionals for laboratory tests. However, legislation at the state level does not address the possibility of obtaining DTC testing in other states.

Yet another approach is to focus on disseminating genomic education toward consumers and healthcare providers regarding best practices in clinical genetic testing. This approach would entail providing consumers with evidence-based information on the degree of the clinical utility of the test, the potential for increasing health-care costs, and options to seek out professional genetic testing services by licensed health-care professionals who can provide interpretation of the results and develop actionable interventions in concert with the patient’s health status, lifestyle, and familial risk factors. Dissemination of genetic education would also include increasing health-care provider knowledge about the array of DTC tests available to consumers and best practices for addressing DTC test results in clinical practice. For many years, the National Institute of Nursing Research, American Nurses Association, and other professional nursing organizations have been refining genomic competencies for nurses to prepare the nursing workforce to work one-on-one with patients and families on issues concerning genetic testing, and as advocates for disseminating information to consumers about their rights and safeguards for preserving the confidentiality of their genetic data.

The Academy’s Position

The American Academy of Nursing has long advocated for all patients’ rights to information, education, and privacy related to their health care. As genetics has begun to play an increasingly important role in the provision of clinical care and research, it is imperative that patients are aware of the dangers and clinical limits of DTC genetic testing, and that appropriate information is provided for interpretation of the results based on the context of the individual’s health, lifestyle, and family history. Unchecked, these predatory companies will continue to provide testing to consumers with unknown analytic and clinical validity and clinical utility, and have a high potential to undermine the scientifically supported application of genetics in clinical settings. Further, individuals are at high risk of future discrimination for genetic findings based on these unregulated tests.

For these reasons, the American Academy of Nursing supports efforts to increase the FDA’s regulation of private companies offering DTC genetic testing, improve transparent marketing to consumers, develop policy to protect consumers from discrimination and unnecessary health-care costs based on genetic information acquired through DTC testing, and educate nurses about best practices for genetic testing and clinical utility of genetic test results, as well as to increase nurses’ role in genetic education for patients and families.

Recommendations

The Academy will collaborate with organizations, including the American Nurses Association, the American Medical Association, the American Association of Colleges of Nursing, the American Society of Human Genetics, the National Institute of Nursing Research, and the International Society of Nurses in Genetics, and other clinician and consumer organizations, to educate legislators and the general public about best practices for genetic testing to put the patient’s best interests and the public’s health ahead of revenue-focused, private companies. The Academy urges the CMS and FDA to work with entities that own and market LDT and DTC genetic tests and services regarding the clarity of the information that is provided to consumers. This information includes

(1) premarket demonstration of test validity and reliability, and coordination with the FTC to ensure that the advertisements accurately reflect the information that will be given to consumers;
(2) clinical utility of the test, accessible explanation of the test results, potential breach of privacy, and loss of confidentiality;
(3) alternative options for testing and how to access consultation with a licensed genetic counselor or other health-care professionals to assist with interpretation and options; and
(4) disclosure of potential for increasing out-of-pocket expenses for repeat testing and/or office visits, and/or interventions.

The Academy requests clarification by the CMS regarding coverage of retesting when genetic test results are furnished from LDT and DTC entities, along with recommendations for reducing unnecessary downstream interventions while preserving patient safety and quality of care.

REFERENCES