

ASHG Statement* on Direct-to-Consumer Genetic Testing in the United States

Direct-to-consumer (DTC) genetic testing has been gaining prominence over the past several years.¹ Proponents of DTC testing cite benefits that include increased consumer access to testing, greater consumer autonomy and empowerment, and enhanced privacy of the information obtained. Critics of DTC genetic testing have pointed to the risks that consumers will choose testing without adequate context or counseling, will receive tests from laboratories of dubious quality, and will be misled by unproven claims of benefit.

Currently, DTC genetic testing is permitted in about half the states² and is subject to little oversight at the federal level. In July 2006, the Government Accountability Office issued a report documenting troubling marketing practices by some DTC testing companies,³ and the Federal Trade Commission (FTC) issued a consumer alert cautioning consumers to be skeptical about claims made by some DTC companies.⁴ Internationally, several countries have issued reports cautioning against its use,⁵⁻⁷ and several European countries have banned or are considering banning it entirely.

DTC testing has emerged during a period of rapid growth in the number of genetic tests. Today, there are more than 1,100 genetic tests available clinically, and several hundred more are available in research settings. Although most genetic testing is currently available only through a health care provider, an increasing variety of tests are being offered DTC, often without any health care provider involvement or counseling. The range of tests available DTC is broad, from tests for single-gene disorders, such as cystic fibrosis, to tests for predisposition to complex, multifactorial diseases, such as depression and cardiovascular disease. In addition to providing test results DTC, some companies also make recommendations regarding lifestyle changes on the basis of these results, such as changes in diet or use of nutritional supplements.

Ensuring adequate information, high-quality laboratories, and accurate claims and interpretation of test results is important for all genetic tests, including those provided DTC. At the same time, a one-size-fits-all approach is not appropriate for DTC tests, because the types of tests being offered are heterogeneous, and their consequences are wide ranging. A test may be used to diagnose disease, to predict risk of future disease, to determine the risk of passing on a disease to one's offspring, to aid in therapy selection, or to guide "lifestyle" choices such as diet and skin care. Different possible actions may result from different types of tests. For example, tests to determine whether someone is a carrier of a mutation for a particular disease may affect the choice of whether or whom to marry, whether to have children, and whether to terminate a pregnancy. Thus, the level of evidence required before a test is offered DTC, and the safeguards appropriate to ensure adequate consumer protection, will differ depending on what is being tested for and what the foreseeable consequences of testing are. Whereas the DTC model may be contraindicated for certain types of tests, the availability of other tests in the absence of a

health care provider may not compromise, and may even foster, patient health. This policy statement does not attempt to set the dividing line between those tests that should be offered DTC and those that should not; rather, it sets forth principles that should govern all health-related genetic tests that are offered DTC.

Scope of this Statement

While DTC testing also encompasses paternity and ancestry testing, this policy statement addresses solely those genetic tests that make health-related claims or that directly affect health care decision making. In addition, although "DTC" is sometimes used to refer to tests advertised but not sold DTC, this policy statement focuses on tests that can be ordered directly by a consumer and whose results are reported DTC without an independent health care provider—one not employed by the testing company—serving as an intermediary.

Context

DTC genetic testing differs from traditional genetic testing in that consumers order tests and receive test results without an independent provider serving as an intermediary. Whether a company is permitted to provide DTC genetic-testing services is a matter of state law. Currently, about half the states permit DTC genetic testing.² Additionally, although some states require a provider to order a test on behalf of a patient, this requirement can generally be fulfilled by a physician employed by the laboratory. Some DTC companies offer genetic counseling, while others do not.

DTC tests are typically advertised and sold over the Internet. After the consumer orders the test, the testing company sends a sample-collection kit (e.g., buccal swab or blood-spot collection). The consumer sends back the sample, and the company performs the test and sends a test report via the Internet or the mail. This context has led to the concern that consumers will not receive adequate counseling—either in advance, to ensure that the test is appropriate, or on receipt of test results, to ensure that consumers comprehend the complex information and understand the consequences of testing for themselves and their family members.

Quality

Because of the fragmented regulatory environment for genetic testing in general, there is concern that the quality of the tests offered DTC may be inadequate. For a test to be of good quality, the laboratory performing it must be able to obtain the correct answer reliably, meaning that it detects a particular genetic variant when it is present and does not detect the variant when it is absent. A test's accuracy is referred to as "analytic validity." Further, there must be adequate scientific evidence to support the

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correlation between the genetic variant and a particular health condition or risk—the so-called clinical validity.

Currently, the federal government exercises limited oversight of the analytic validity of genetic tests and virtually no oversight of their clinical validity. Laboratories that perform clinical genetic testing must be certified under the Clinical Laboratory Improvement Amendments of 1988 (CLIA). However, although CLIA imposes basic requirements that address personnel qualifications, quality-control standards, and documentation and validation of tests and procedures, it does not address clinical validity or claims made by the laboratory regarding the tests. Nor does CLIA yet contain a “specialty area” for most genetic tests, which hampers the government’s ability to determine whether tests are being performed correctly.⁸ Although the Centers for Medicare and Medicaid Services (CMS) stated for several years that it intended to create a genetic-testing specialty, the agency suddenly reversed course in 2006, stating that no specialty would be issued.

The level of scrutiny by the U.S. Food and Drug Administration (FDA) differs markedly depending on whether the test is performed using a commercial “test kit” or a laboratory-developed test method. Whereas the FDA reviews the analytic and clinical validity and the labeling of commercial test kits before they are marketed and requires postmarket adverse-event reporting if there are problems with the kit, there is no premarket review of laboratory-developed tests, nor is there any requirement to report adverse events. Recently, the FDA indicated that it plans to regulate a small subset of laboratory-developed tests known as “in vitro diagnostic multivariate index assays,”⁹ but this is a very narrow category of tests that will exclude the vast majority of genetic tests offered by clinical laboratories.

The lack of a coherent regulatory landscape to ensure quality is not unique to DTC genetic testing, since all other molecular and biochemical tests are also affected. However, quality concerns are particularly acute in the DTC context because of the low barrier to market entry, the complexity of the information that consumers need to understand to make an informed decision, and the lack of provider scrutiny. Consumers are at a significant risk of selecting tests with unproven benefit, of obtaining testing services from laboratories of dubious quality, and of making decisions without timely and accurate genetic counseling.

Claims

Claims made regarding DTC genetic tests may in some cases be exaggerated or unsupported by scientific evidence. Exaggerated or unsupported claims may lead consumers to get tested inappropriately or to have false expectations regarding the benefits of testing. Further, consumers may make unwarranted, and even irrevocable, decisions on the basis of test results and associated information, such as the decision to terminate a pregnancy, to forgo needed treatment, or to pursue unproven therapies.

Some DTC companies use privacy as a marketing tool, touting the benefits of obtaining genetic testing outside the health care system and thereby avoiding the risks of having genetic information contained in a medical record. However, these companies do not necessarily disclose their privacy policies or explain that a patient’s subsequent disclosure of the test results to a physician may lead to the information becoming part of his or her medical record. Further, DTC companies are not necessarily subject to the health privacy regulations issued pursuant to the Health Insurance Portability and Accountability Act (HIPAA), leaving consumers vulnerable to having their information used or disclosed in a manner that would be impermissible in the health care system.

Federal law prohibits companies from using unfair, deceptive, or fraudulent trade practices, including making false or misleading advertising claims. This law, in theory, prohibits clearly false genetic-testing claims. Several complaints have been filed and are pending with the FTC about a specific DTC genetic-testing company, and the FTC recently issued a consumer alert warning the public that “some of these [DTC] tests lack scientific validity, and others provide medical results that are meaningful only in the context of a full medical evaluation.”⁴ The FTC has not, however, taken direct action against any DTC genetic-testing company. Furthermore, with respect to tests for which some scientific support exists but for which claims are exaggerated or provide incomplete information, FTC regulators may be insufficiently knowledgeable to detect the misleading nature of such claims. It also must be recognized that there are limits to the government’s ability to restrict commercial speech. Finally, although the FDA has the authority to regulate claims for products it regulates, the agency currently does not regulate most genetic tests and therefore does not regulate their claims.

On the basis of the foregoing analysis, the American Society of Human Genetics makes the following recommendations about DTC genetic testing.

Recommendations

I. Transparency

To promote transparency and to permit providers and consumers to make informed decisions about DTC genetic testing, companies must provide all relevant information about offered tests in a readily accessible and understandable manner.

- a. Companies offering DTC genetic testing should disclose the sensitivity, specificity, and predictive value of the test, and the populations for which this information is known, in a readily understandable and accessible fashion.
- b. Companies offering DTC testing should disclose the strength of scientific evidence on which any claims of benefit are based, as well as any limitations to the claimed benefits. For example, if a disease or condition may be caused by many factors, including the presence of a particular genetic variant, the company should disclose that other factors may cause the condition and that absence of the variant does not mean the patient is not at risk for the disease.
- c. Companies offering DTC testing should clearly disclose all risks associated with testing, including psychological risks and risks to family members.
- d. Companies offering DTC testing should disclose the CLIA certification status of the laboratory performing the genetic testing.
- e. Companies offering DTC testing should maintain the privacy of all genetic information and disclose their privacy policies, including whether they comply with HIPAA.
- f. Companies offering DTC testing and making lifestyle, nutritional, pharmacologic, or other treatment recommendations on the basis of the results of those tests should disclose the clinical evidence for and against the efficacy of such interventions, with respect to those specific recommendations and indications.

II. Provider Education

To ensure that providers are aware that genetic tests are being provided DTC and that some of these tests may lack analytic or clinical validity,

professional organizations should educate their members regarding the types of genetic tests offered DTC, so that providers can counsel their patients about the potential value and limitations of DTC testing.

- a. Professional organizations should disseminate information to their members explaining what DTC testing is, what tests are offered DTC, and the potential benefits and limitations of such testing for patients.

III. Test and Laboratory Quality

To ensure the analytic and clinical validity of genetic tests offered DTC and to ensure that claims made about these tests are truthful and not misleading, the relevant agencies of the federal government should take appropriate and targeted regulatory action.

- a. CMS should create a genetic-testing specialty under CLIA, to ensure the analytic validity of tests and the quality of genetic-testing laboratories.
- b. CMS should ensure that all DTC genetic-testing laboratories are certified under CLIA and should maintain a publicly accessible list containing the certification status of laboratories.
- c. The federal government should take steps to ensure the clinical validity of DTC tests that make health-related or health care-affecting claims.
- d. The FTC should take action against companies that make false or misleading claims about DTC tests.
- e. The FDA and the FTC should work together to develop guidelines for DTC testing companies to follow, to ensure that their claims are truthful and not misleading and that they adequately convey the scientific limitations for particular tests.
- f. The Centers for Disease Control and Prevention (CDC) should conduct a study on the impact of DTC testing on consumers, to assess whether and to what extent consumers are experiencing benefit and/or harm from this method of test delivery. The CDC should also conduct a systematic comparison between the claims made in DTC advertising and the scientific evidence available to support these claims.

Conclusion

DTC genetic testing is a method of marketing genetic tests to consumers without the involvement of an independent health care provider. Potential benefits of DTC testing include increased consumer awareness of and access to testing. In the current environment, consumers are at risk of harm from DTC testing if testing is performed by laboratories that are not of high quality, if tests lack adequate analytic or clinical validity, if claims made about tests are false or misleading, and if inadequate information and counseling are provided to permit the consumer to make an informed decision about whether testing is appropriate and about what actions to take on the basis of test results.

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COMMITTEE

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