



Direct-to-Consumer Genetic Testing

What is a Direct-to-Consumer genetic test?

Direct-to-Consumer (DTC) genetic tests, also known as Personal Genomic Profiles or Informational Genetic Tests, are genetic tests marketed and sold directly to consumers, with subsequent test results available directly to the purchasing consumer. DTC genetic tests do not require the assistance of a physician or other health care provider to obtain or interpret. DTC genetic tests cover a broad range of types: carrier testing for diseases such as cystic fibrosis and hemochromatosis; prenatal testing; newborn screening; pharmacogenomic testing; diagnostic testing; testing for predisposition to complex diseases such as hereditary cancers, cardiovascular disease, and depression; and testing to determine ancestry.

Where do my patients obtain DTC genetic tests?

Several companies offer DTC genetic testing, mainly through the internet. Consumers can purchase a specimen collection kit online. When the kit arrives, the consumer collects the specimen, often saliva or a cheek swab, and then mails it back to the company. The test is performed, and then results are posted on a secure website that only the purchaser (and the company) can access.

What do the test results mean?

The results of a genetic tests (whether DTC or ordered by a physician) can be challenging to interpret. A positive result does not necessarily indicate a clinical diagnosis. Often, a positive result indicates an increased risk for developing a disease or condition. Phenotypic manifestations are also different in individuals. The same mutation in different people can have different penetrance and expressivity, and can be influenced by different environmental factors. Also, since only a fraction of testable mutations are identified for genetically based diseases, a genetic test with a negative result is not indicative of the absence of disease risk. A patient receiving such a result either has the same risk of developing the disease as the general population, or may carry a mutation that has not yet been identified that confers increased disease risk. These concepts are seldom, if ever, communicated to consumers by companies marketing DTC genetic testing, and may be difficult for physicians to explain to patients. A few companies offering DTC genetic testing include the services of a genetic counselor or other genetics professional to assist consumer and physicians in interpreting DTC test results.

The results of some DTC genetic tests may aid in treatment decisions. For example, a test result indicating a patient has an increased risk for cardiovascular disease may spur a recommendation that your patient lead a healthy lifestyle by exercising regularly and

eating healthfully. However, genetic test results, particularly those pertaining to complex diseases, must be interpreted in the context of the patient's other health factors (e.g., family history, environmental characteristics, other health conditions, current medications, etc.)

What do I say to a patient if he or she is interested in obtaining DTC genetic testing?

AMA policy encourages patients to undergo genetic testing under the guidance of a qualified health care professional. Patients will benefit from a discussion about the benefits and risks of DTC genetic testing, and need guidance in determining whether particular tests are appropriate for them. If a physician feels unprepared to have such a discussion, referral to a clinical geneticist or a genetic counselor is encouraged.