

## Direct-to-consumer genetic testing

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### Types of DTC genetic tests

A broad range of genetic test types are offered DTC, including:

- Carrier testing for diseases such as cystic fibrosis and hemochromatosis
- Pharmacogenomic testing
- Testing for predisposition to complex diseases such as hereditary cancers, cardiovascular disease and depression
- Whole exome or genome sequencing
- Testing to determine ancestry

### Obtaining a DTC genetic test

Several companies offer DTC genetic testing, mainly through the internet. Consumers can purchase a specimen collection kit online, and 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. Most companies offering DTC genetic testing will ask the consumer to consent to using his or her genetic data for further studies.

### Interpreting results of DTC genetic tests

The results of genetic tests (whether DTC or ordered by a physician) can be challenging to interpret. A positive result does not always indicate a clinical diagnosis. Instead, it may indicate an increased risk for developing a disease or condition.

Similarly, a negative result is not indicative of the absence of disease risk. These concepts can be difficult for consumers to understand without a physician or genetic counselor to fully explain them.

Genetic test results, particularly those pertaining to complex diseases, must be interpreted in the context of the patient's other health factors, like family history, environmental characteristics, other

health conditions and current medications.

## **Patients interested in DTC genetic testing**

AMA policy encourages patients to undergo genetic testing under the guidance of a physician or genetic counselor. Physicians should discuss with their patients the benefits and limitations of DTC genetic testing, and provide guidance in determining whether particular tests are appropriate for them. Referral to a clinical geneticist or a genetic counselor is encouraged if physicians are not able to answer detailed questions about DTC genetic testing.

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