

Connecticut Department of Public Health  
Genomics Office

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**DIRECT-TO-CONSUMER PERSONAL GENOMICS SERVICES  
INFORMATION TO CONSIDER**

*It is important to recognize that factors other than genes, including diet, exercise, and environmental exposures, also contribute to disease.*

**INTRODUCTION**

Genetic testing has rapidly moved forward since the human genome was decoded in 2003. Previously the use of genetic testing was limited primarily to conditions, such as sickle cell anemia, cystic fibrosis, and Huntington's disease, caused by genetic changes in single genes. New technology allows the simultaneous analyses of hundreds of thousands of genes in an attempt to find the underlying genetic causes of more complex common diseases like cancer and diabetes.

Embracing the advancements in genetic and Internet technology, a number of web-based companies have started offering personal genome profiles directly to consumers. The companies claim to provide consumers with information about their risk of developing diseases such as asthma, diabetes, heart disease, and various cancers. The presumed goal is to empower individuals to take better control of their health. Some companies also offer direct-to-consumer (DTC) genetic testing for purposes of determining ancestry or to help individuals personalize their diets.

The information that follows focuses primarily on DTC genetic testing that is being offered to provide you with information about your chances of developing specific diseases in the future.

Although genetic tests have traditionally been available only through healthcare providers, you can now order a test-kit directly through the mail. The test typically involves collecting a saliva sample at home and mailing it back to the company's laboratory. A few weeks later, your results are available electronically on a secure website with information about your chances of developing specific diseases or conditions. If you have questions about your results, you can usually contact a genetic counselor made available through the testing company. The price for this type of at-home testing ranges from several hundred dollars to thousands of dollars.

Although the testing process may seem simple, interpreting the results is not straightforward. It is often unclear what you should do with the information you get. For example, suppose that your test result indicates that you have a 20 percent increased risk of developing a particular disease in your lifetime. Is this enough for you to be concerned and to seek follow-up care? How does this compare with the risk in the general population? Will your doctor know what to do with the information, should you decide to share it with him/her? How does this information fit in with your personal and family health history or with other factors, such as diet, exercise, and environmental exposures (e.g., air pollution, tobacco smoke, pesticides) that may also contribute to the development of the disease? Therefore, it is important to ask yourself the following questions:

- What information do I hope to get from these tests?
- Will the information be useful?
- Can my disease risk change over time?
- What will I do with the information I receive?
- What happens with my sample and genetic information after genetic testing has been completed?

Besides the difficulty of making sense out of genetic test results, it is also important to be aware that genetic testing has many other limitations.

## **GENETIC VARIATION**

Although most of your DNA is the same as any other unrelated person's DNA, small differences exist. This genetic variation makes up about one-tenth of a percent of your DNA and is what makes your physical appearance and health unique. Some variations in DNA involve whole segments while other differences involve only a single unit or base. Locations in the DNA sequence where individuals differ at a single DNA base are called single nucleotide polymorphisms, or SNPs (pronounced "snips"). SNPs are not necessarily located in the genes that directly cause a disease. They may be located in a piece of DNA that is close to the disease gene and may be inherited along with the gene. Therefore, the detection of the SNP through genetic testing may serve as a marker that the disease-associated gene is also present.

Genetic testing offered directly to consumers involves examining a person's genetic material to see if any differences exist that may indicate a person has a greater than average risk of

developing particular diseases. These tests are based on studies that indicate certain SNPs have been found to be associated with the development of specific diseases. Although genetic testing has been around for years, most new commercial genetic tests use what is known as DNA microarray technology to evaluate several hundred thousand SNPs at a time rather than just a few specific genes. It is unclear, however, just how useful the information gained from SNP analysis really is. For instance, even if it is found that a person has several SNPs that suggest an increased risk of getting a disease, that person may have many other SNPs, yet undiscovered, that indicate a lower risk.

Although SNPs have been the most widely studied type of genetic variation and form the basis of most of the commercially available DTC genetic tests, the results of these efforts have not yet explained much of the heritability of disease,<sup>1</sup> thus bringing into question their usefulness. Studies surrounding other types of genetic variation and disease-association are still in their infancy, but ultimately will increase our understanding of disease susceptibility.

## **LIMITATIONS OF GENETIC TESTING FOR INHERITED SUSCEPTIBILITY TO COMMON DISEASES**

Although commercial genetic tests may sound appealing and harmless, it is important to be aware of some of the pitfalls and limitations of such tests.

**Results from genetic testing are not necessarily “black and white” – there remains uncertainty.** Genetic information is complex. The genetic tests that you can order directly online do not give you a simple ‘yes’ or ‘no’ answer as to whether you will get a particular disease during your lifetime. They only tell you the chances that you will develop a particular disease and only to the extent that the SNPs tested contribute to your risk for a particular disease. Even if a test reveals that you have a high probability of developing a certain disease, it does not tell you when you might get the disease or how severe the symptoms of the disease will be. On the other hand, not being identified as having a particular genetic susceptibility does not mean that you will not develop the disease. In addition, many factors other than genes, including diet, exercise, and environmental exposures also contribute to your health status.

**The number of SNPs tested for is limited.** Direct-to-consumer genetic tests based on SNPs assess only a limited number of gene variants found to be associated with any given disease. Many rare variants remain to be discovered. Thus you may have many SNPs, yet

undiscovered, that could change your risk status from above to below average risk of disease, or vice versa. Inconsistent information about your risk status over time may be problematic if follow-up recommendations and the need for lifestyle changes vary accordingly.<sup>2</sup>

**Most SNPs discovered so far have only a small effect on disease risk.** SNPs that have been identified account for only a small fraction of the genetic variation among individuals.<sup>3</sup> Other sources of genetic variation that could affect your risk for disease include variation in the number of copies of genes that you have and differences in how genes are expressed. It is possible that some of these types of genetic variation may be found to have a greater effect on disease risk than SNPs, but more studies are needed to investigate their contribution. Future genetic tests may incorporate these other types of genetic variation in assessing a person's risk.

**Direct-to-consumer genetic testing may not be worth the out-of-pocket cost.** The use of currently known genetic variants in predicting the development of common diseases may not add substantially to risk prediction achieved through an assessment of family health history or established risk factors, such as high cholesterol levels or high blood pressure.<sup>4</sup> Therefore, the value added by DTC genetic testing may not be worth the cost at this time.

**There is a lack of standardization among consumer genomics companies in a number of key areas.** Your personal genome profile may differ depending on which company you choose to do the testing. Consumer genomics companies tend to differ on (1) the number of diseases for which you can be tested; (2) the scientific studies they consider when deciding which SNPs to use for testing; (3) the type and number of SNPs they use to calculate genetic risk for a particular disease; and (4) the statistical methods they use to estimate risk. There have been instances when DNA samples from the same person were sent to different companies, resulting in inconsistent and misleading results.<sup>5</sup>

**Government oversight of DTC genetic testing is limited.** There is no regulatory system to ensure that this type of genetic testing is supported by adequate evidence before being marketed, or that marketing claims for such tests are truthful and not misleading. This raises questions about the validity as well as the usefulness of some of these DTC genetic tests.

**Results may not apply to all ethnicities.** Direct-to-consumer companies get most of their information from studies based upon disease associations in people of European descent.

Therefore, some of the risk information that these companies provide may not apply to people coming from other ethnic backgrounds.

**Genetic counseling may be needed.** Although many of the consumer genomics companies have genetic counselors available for counseling by phone, you are basically on your own to interpret your results, which may not be easily understood or which may cause you and your family to worry unnecessarily. Therefore, it is important to consider alternatives such as genetic testing in a clinical setting that includes a consultation with a genetic counselor or knowledgeable healthcare provider before and after testing. A trained genetic counselor can help you consider the emotional aspects of genetic testing and understand what it may, or may not, tell you. After testing, your counselor can help you interpret the results and their relevance to you and your family.

## **PRIVACY ISSUES**

The federal Genetic Information Nondiscrimination Act (GINA), passed in May 2008, provides protection against discrimination in health insurance and employment based on a person's genetic information. However, you should not assume that federal or state health privacy laws offer you privacy protection regarding the maintenance, use, or disclosure of your genetic information if a commercial company obtains health information about you. When considering genetic testing, you should be informed about the company's policies and practices regarding the following issues:

- **Storage issues related to genetic material after sample collection.** How long are DNA samples kept? How securely are samples kept? How are the samples destroyed? Are the samples available to researchers or others?
- **Access issues related to your genetic information.** Who has access to your information? How is it protected? Can the company sell information about you to others even if it is made anonymous?

## **PROFESSIONAL ORGANIZATION STATEMENTS**

The National Society of Genetic Counselors, the American Society of Human Genetics, and the American College of Medical Genetics have all issued statements about direct-to-consumer genetic testing. They recommend that companies provide a clear statement of scientific evidence for a test, that testing labs be appropriately certified, that there is the involvement of a

genetics expert to order and interpret the test, and that consumers should be fully informed about the meaning of test results and the security measures in place to protect the privacy of their results.

## **GENETIC TESTING AND PUBLIC HEALTH**

With an ever-growing number of genetic tests being developed to identify a person's likelihood of developing common chronic diseases or increased susceptibility to specific infectious diseases, comes the potential for prevention and improvement in the public's health.

For example, one study from the Diabetes and Prevention Program of the National Institute of Diabetes and Digestive and Kidney Diseases found that those genetically predisposed to diabetes could delay or in some cases even prevent the disease from developing by making lifestyle changes, such as eating fewer calories, getting calories from the right kinds of foods, and exercising more than two hours a week.<sup>6</sup> Therefore, genetic screening programs that can identify individuals at high risk of a disease, like diabetes, may be better able to prevent disease progression and help determine the most appropriate treatments. However, it is critical that genetic tests are accurate, reliable, safe, useful, and affordable if the public is going to rely on them.

## ADDITIONAL LINKS AND RESOURCES

[Human Genetic Variation Fact Sheet](#), National Institute of General Medical Sciences, July 2008.

[Statement on Direct-to-Consumer Genetic Testing](#), American College of Medical Genetics, 2008.

[Statement on Direct-to-Consumer Genetic Testing in the United States](#), American Society of Human Genetics, September 2007.

[Position Statement on Direct-to-Consumer Genetic Testing](#), National Society of Genetic Counselors, 2007.

[At-Home Genetic Tests: A Healthy Dose of Skepticism May Be the Best Prescription](#), Federal Trade Commission, July 2006.

[Promotion of Genetic Testing Services Directly to Consumers](#), Genetic Alliance.

[Issue Brief on Direct-to-Consumer Genetic Testing: Empowering or Endangering the Public?](#) Genetics and Public Policy Center, July 2006.

## REFERENCES

- 1 Maher B. Personal genomes: The case of the missing heritability. *Nature* 2008;,456:18-21.
- 2 Mihaescu R, van Hoek M, Sijbrands EJG, et al. Evaluation of risk prediction updates from commercial genome-wide scans. *Genet Med* 2009;11:588-594.
- 3 Kraft P, Hunter DJ. Genetic risk prediction – Are we there yet? *N Engl J Med* 2009;360:1701-1703.
- 4 Meigs JB, Shrader P, Sullivan LM, et al. Genotype score in addition to common risk factors for prediction of type 2 diabetes. *N Engl J Med* 2008;359:2208-2219.
- 5 Ng PC, Murray SS, Levy S, Venter JC. An agenda for personalized medicine. *Nature* 2009;461:724-726.
- 6 <http://diabetes.niddk.nih.gov/dm/pubs/preventionprogram/index.htm>. Accessed on July 2, 2009.

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