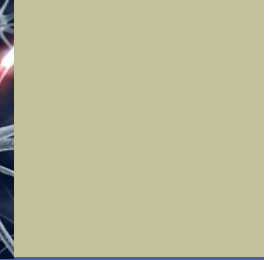
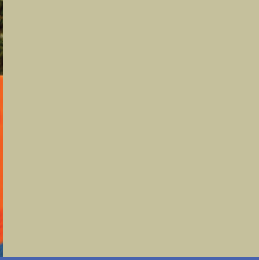
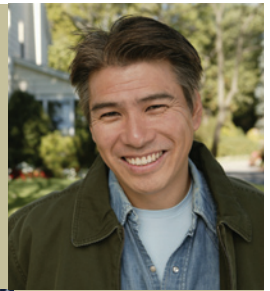


NATIONAL  
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# Looking at My Genes: What Can They Tell Me?

Frequently Asked Questions About  
Genome Scans and Genetic Testing

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U.S. Department of Health  
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# Can Gene Scans Tell Me About My Risk of Mental Illnesses?

New tests that scan all of a person's genes – that person's "genome" – or large parts of his or her genome are now on the market. Anyone who can afford the new scans can buy one, without a prescription or a health professional's advice, by mailing a saliva sample to a company that sells the scans. Advertisements suggest that the company then can provide information about clients' risks of developing specific diseases, based on variations found in their genes.

It's too early for these new genome scans to give people a complete picture of their risk of mental illnesses or to diagnose them. Scientists don't yet know all of the gene variations that contribute to mental illnesses, and those that are known, so far, raise the risk by very small amounts.

Genetic research might make it possible – one day – to provide a more complete picture of a person's risk of getting a particular kind of mental illness or to diagnose it, based on his or her genes. That day isn't here yet, although studies are underway now.

In the meantime, it's important to know the difference between the new genome scans being advertised today and the kinds of traditional genetic tests doctors have been ordering for many years. Traditional genetic tests look for specific variations **clearly shown** by research **to be the cause of** certain rare diseases, such as cystic fibrosis, that generally are caused by variations in a single gene.

Doctors order traditional genetic testing for people they think are at high risk of one of these rare diseases; for example, if the neurological disorder Huntington's disease runs in a person's family. The results enable patients and their doctors to make health care decisions together.

But people who buy the new genome scans may not have any reason, such as family history, to suspect they're at risk of one of these rare

diseases. They may just want to randomly look at all of their genes with the hope of finding out if they're at risk of **any** diseases. As noted, when it comes to common diseases, like mental illness or adult onset diabetes, not enough is known about which gene variations are involved to give a complete picture of a person's risk.

Also, the genetics of mental illnesses and other common diseases are much more complex than the genetics of many of the rare, single-gene diseases. Mental illnesses appear to involve variations in many genes combined with other factors, such as stress.

People who are thinking about buying one of the new genome scans, to look at all or most of their genes with the hope of finding out if they're at risk of a mental illness or another common disease, may want to get their health-care providers' advice before taking that step.

People who suspect they have a rare disease strongly tied to their genetic make up – for example, if a rare disease runs in their family – may want to ask their health care providers about genetic testing. Their providers can tell them whether or not the disease can be detected through genetic testing at this time and, if so, what kind of test and follow-up care are needed.

## Q. What are the new genome scans that recently appeared on the market?

**A.** In recent years, scientists identified almost all of the genes in humans and the most common variations in many of them. This has proven to be a valuable tool for research.

Some companies are using this new information to market genome scans directly to consumers, which anyone can buy without a doctor's advice. Clients send their saliva to the company, and the company examines the saliva for variations in the clients' genes.

The company tells the client what gene variations he or she has, and offers to analyze the results or to allow the client to analyze the results on an interactive Web site.

## Q. Can the new genome scans tell me what diseases I might get?

**A.** To date, no gene variants are known that can predict with certainty whether or not someone will get a number of common diseases, including mental illnesses. Scientists haven't yet discovered whether many of the gene variations that occur in humans are connected to specific diseases or how much they raise or lower the risk.

For example, a genome scan might show that a person has one or more gene variations related to a common disease, such as adult-onset diabetes. But it's likely that many other gene variations also contribute to this disease, and it's not yet known which ones. The variations associated with most common diseases, to date, raise the risk only very slightly and, by themselves, don't yet provide medically useful information.

An example using heart disease suggests how the results of the new genome scans might confuse consumers. Early research might have shown that certain gene variations are associated with a common kind of heart disease. People whose scans showed they **didn't** have these variations might think they weren't at risk.

They might think it would be safer for them than for other people to forego some of the healthiest, heart-protecting habits known to science: exercising, not smoking, and avoiding obesity. But it's likely that, as with mental illnesses, this kind of heart disease involves many variations in many genes, and scientists don't yet know what all of them are. Foregoing healthy lifestyle changes based on this incomplete genetic picture could contribute to illness that might have been prevented.

## Q. What is traditional genetic testing?

**A.** Like the new genome scans being marketed to consumers, traditional genetic testing looks for variations in a person's genes by examining body fluids or tissues. A major difference is that traditional genetic tests focus on specific variations in a single gene **known with certainty** to be the cause of a rare disease, and genetic testing usually is ordered by a doctor working closely with a patient.

Doctors are looking for evidence of a specific disease when they order traditional genetic testing; for example, if they know that a disease runs in a person's family.

## Q. Can traditional genetic testing tell me what diseases I might get?

**A.** Genetic tests for some rare diseases clearly tied to a specific, single gene, like cystic fibrosis, fragile X syndrome (a heritable cause of mental retardation), or sickle cell disease, give people definitive answers about their risk of getting these rare illnesses. Doctors have ordered genetic tests for these kinds of illnesses for many years.

The results have given people valuable health information that has helped them get the right treatments or, if no treatment is available, to plan their lives and care, in consultation with their health-care team.

## Q. Should I have traditional genetic testing?

**A.** Your health-care provider can help you decide whether to have traditional genetic testing; for example, if you know that a rare disease with a clearly identified genetic basis runs in your family, and you want to know if your parents passed on to you the gene variations that cause the disease – and whether you're at risk of passing the variations to your children. Your provider can determine exactly which test is needed and can arrange genetic counseling – an important part of genetic testing, described in the next section.

Test results may put your mind at ease if they show you don't carry the genetic variation that causes the disease. If the results show that you do have the variation, that may help you make life plans, such as whether to have children or adopt. Your health-care team will play a key role, interpreting the test results for you and helping you decide how the results will affect important life decisions.

Some rare genetic diseases, or their symptoms, can be prevented or treated, and genetic testing can help you and your health-care provider take action. If there is no effective treatment, you and your provider can take measures to improve your health and your life.

## Q. What is genetic counseling?

**A.** Genetic counseling provides information and support to people who have, or may be at risk of, inherited disorders. A genetics professional (medical geneticist, genetics counselor, or genetics nurse) discusses your risk with you and may or may not suggest genetic testing.

Genetics professionals:

- Assess your risk by looking at your family's health history and medical records.
- Provide support and information to help you decide whether to be tested.
- Explain what the results of genetic tests mean.
- Provide counseling or refer you to support services.
- Explain options for treatment or prevention.
- Explain how test results could affect decisions about having children.

If your test shows that you do have a genetic variation clearly associated with a specific disease, you may be referred to a doctor or researcher who specializes in that illness. Both your primary care provider and a genetics professional can help you with decisions about testing and can help you interpret your test results.

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## Family History Gives You Good Clues About Your Risk

Your family history is one of your best clues about your risk of developing many common illnesses, including mental disorders.

For example, bipolar disorder and schizophrenia tend to run in families. At this time, **no** type of genetic testing can tell you whether or not you will develop mental illnesses. Not enough is known about which gene variations contribute to them, or the degree to which other factors contribute.

For now, your family history may be your best indicator. For example, studies show that if you have a close relative with bipolar disorder, you have about a 10 percent chance of getting a mood disorder, such as bipolar disorder or depression.<sup>1</sup>

Now consider what the gene variations scientists have linked to mental disorders, so far, can tell you about your risk. Even the variations with the strongest ties raise the risk by only very small amounts. Knowing that you have one of these variations won't tell you nearly as much about your risk as your family history can.

Family history also provides a good clue about your risk of rare diseases that **can** be detected through genetic testing. If one of these rare diseases, such as cystic fibrosis, runs in your family, your risk is likely very high.

If a disease runs in your family, your health-care provider can tell you if it's the kind of illness that can be detected through genetic testing at this time. Your provider can help you make decisions about whether to be tested and can help you understand test results and their implications.

<sup>1</sup> Gershon ES, Hamovit J, Guroff JJ, Dibble E, Leckman JF, Sceery W, Targum SD, Nurnberger JI Jr, Goldin LR, Bunney WE Jr. A family study of schizoaffective, bipolar I, bipolar II, unipolar, and normal control probands. *Arch Gen Psychiatry*. 1982 Oct;39(10):1157-67. PMID: 7125846.

## What Are Genes?

Genes are pieces of DNA, in cells, that parents pass down to their children at conception. Genes turn on and off throughout life to transmit chemical instructions for making the body's proteins. Proteins are part of what gives us our characteristics; for example, our height, eye color, personality, and our chances of getting specific diseases.

Genes can have slight differences in their structure from person to person, and these variations can cause differences in people's characteristics. Gene variations help explain why members of the same family often look alike and have other characteristics in common, such as certain illnesses – because each child inherits a mixture of both parents' gene variations.

Some gene variations make people more vulnerable to different diseases, and some have protective effects against different diseases. Some rare diseases are caused by variations in a single gene. Most common diseases are caused by a mixture of many gene variations, some unknown, and external factors, such as stress or toxic substances.

Some gene variations appear to have no effect on risk of disease.

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Some genetics professionals specialize in a disease category (such as cancer), age group (such as adolescents), or type of counseling (such as prenatal).

### Q. Are there risks associated with genetic testing?

A. Some people who know they're at risk of a serious disease, because the disease runs in the family, avoid having genetic testing. They're afraid their insurance companies or employers will drop them if tests show they have the gene variation that may lead to the disease. They fear they might have trouble getting insurance or a job.

As of March 2008, efforts are underway to enact federal legislation that would make it illegal to discriminate on the basis of genetic test results.

For the rare, single-gene diseases, genetic testing is important. It leads to early detection and therapy for these diseases.

### Q. What can research about gene variations tell us about disease risk?

A. As they continue to discover combinations of gene variations and external factors that contribute to specific diseases, scientists are building a more complete picture of how to detect which people are at risk of common diseases.

This research also is helping to reveal the biological pathways through which gene variations contribute to disease. Scientists can use this information to design better screening and prevention for specific diseases, and find more precise molecular targets at which to aim as they develop new medications.

For more information about mental illness, including science news about gene-related research on mental illness, visit the National Institute of Mental Health Web site at <http://www.nimh.nih.gov/>.

For information about genetics research, visit the National Human Genome Research Institute Web site at <http://www.genome.gov/>.

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