GENETIC TESTING
A Resource from the American College of Preventive Medicine

A Guide for Patients
Thinking about having your genes tested?

Talk to your doctor first … it’s not as simple as it may seem
It sounds almost too good to be true – send in a swab from your cheek and find out what
diseases you are at risk for, what preventive activities you should be doing and what vitamins and
supplements your body really needs. It’s a new type of gene testing being offered directly to
consumers, often over the internet. Is it really that simple? Can these tests really predict our
future health problems? Are they worthwhile?

Having a genetic test is never as simple as it sounds, even when the doctor recommends one.
But there is a big difference between a genetic test that your doctor advises and one that you
purchase on your own.

Our Mysterious Genes
Our genes are our basic unit of heredity. We have two copies of each gene, one from each
parent. The miracle of genes is in the DNA they contain. This DNA provides instructions to build
every protein in our body. These proteins are the building blocks of our body; they are continually
repaired and replaced. When a new one is needed, the appropriate genes are “turned on” and
their DNA is used to build the new protein. Most genes are the same in all people, but we all
have a few that are slightly different.

Genetic tests use DNA collected from a small sample of body tissue or fluids, such as a swab
from inside the cheek, a drop of blood, saliva or amniotic fluid, or a portion of tumor tissue in
cases of cancer. The sample is sent to a lab that uses a computer to analyze the DNA of specific
genes. The lab provides a report of any DNA variations found. These variations contribute to
our uniqueness, but they also may affect how our body functions. Scientists are intensively
studying the impact gene variations on human health.

We still have a lot to learn about our genes. Until a few years ago, we did not even know how
many genes we had. The Human Genome Project took on the seemingly impossible task of
mapping our complete set of genes, called our genome. The project was completed ahead of
schedule because the scientists found that we have fewer genes than most had suspected –
about 25,000 versus predictions of three to four times that number.

An important result of the project was the development of computer technology to analyze our
genes. This technology has continued to develop. We can now scan thousands of genes at the
same time, and within a few years an individual will likely be able to purchase a scan of his or her
complete genome. This technology has opened the door to a new world of genetic testing.
Understanding the Value of Genetic Testing

Genetic testing is most accurate and most useful when a specific defect, or variation, is being investigated. This is the traditional type of genetic testing that doctors have relied on for years. It has limited, but important uses, such as:

- Diagnosing rare diseases, such as sickle cell anemia, cystic fibrosis or Huntington’s Disease that are caused by a defect in a single gene,
- Screening newborn children for known genetic diseases,
- Assisting reproductive planning by identifying abnormal genes that could be passed on to a child before getting pregnant,
- Assisting with understanding risk or prognosis in a few situations, such as breast cancer, where specific genetic defects have been clearly related to the disease course.

The role and value of gene testing will increase in coming years as more tests are proven to be helpful in patient care. One of the most promising uses will be identifying sensitivities to certain medications to reduce serious side-effects. All of these valuable uses of genetic testing have one thing in common – they are used at the direction of your doctor.

But, in the last few years, the scope of testing has dramatically increased as private companies take advantage of the new technology to offer “gene screens” directly to consumers. These tests can identify differences in genes that seem to occur more often in people with certain common diseases. The implication is that having the specific gene “defect” increases the risk for that disease. However, these relationships are poorly understood. There may be hundreds of variations in genes associated with a specific disease. Some may increase risk; some may not. Some may even be protective. We just don’t understand these effects at this time.

When should a person consider genetic testing?

There are times when your doctor may talk to you about genetic testing. These situations include:

- If you have a family history of an inherited disease, such as muscular dystrophy or sickle cell anemia.
- If you have a strong family history of cancer, especially breast or colon cancer.
- If your parents, brothers or sisters, or other relatives have developed common diseases, such as heart disease or Parkinson’s disease, at a young age (before 50 years).
- If you are a member of an ethnic group with high risk for a particular genetic disease.
- If you are considering pregnancy and you or your partner have a family history of, or are in a high risk group for, an inherited disease.
- If you have a child who has physical or behavioral characteristics suggestive of a genetic syndrome.

Issues to Consider

If you are thinking about having a genetic test, ask your doctor or other health professional the following questions:

- What will this test tell me? Would you recommend that I have it?
- If I do a genetic test on my own, will you go over the results with me?
- Can I trust the result?
- What would a positive result mean? What would you recommend then?
- What is the impact on my family and other relatives? Should I tell them I am being tested? Should they be tested if I am positive?
- How would it affect my health insurance? Would I have to reveal the results?
- What other risks are there with testing? How would I protect my privacy?
Bottom Line
The field of genetic testing is rapidly developing and changing. Two things are for certain. First, genetic tests will be used more and more in medical care as new tests are proven to be effective. There will be more clinical situations when a specific genetic test will aide in decision-making. Second, you will see more and more gene tests that are offered without going through a doctor. These tests will make increasingly appealing claims, from estimating disease risks to recommending optimal nutrition to forecasting your potential. But, at this time, there is little value in these general "do it yourself" gene screens.

Two take home messages: 1) Pay attention to your family history. It is still a reliable source of information about genetic risks. 2) Don't make a decision about having a genetic test alone. Discuss it with your doctor or other licensed health professional. They are still your best advocate when making any decision about medical testing or management.

For other information and useful links, visit the American College of Preventive Medicine website at www.acpm.org.